# Review of the

# National Newborn Screening Programme

# for

# Inherited Metabolic Disorders

**October 2004**

**Programme of Action for Children**

# FOREWORD

The screening of newborn children for metabolic disorders is one of the unsung successes of the health service in Ireland. The screening programme enables potentially devastating diseases to be detected at a very early stage and allow treatment to be started immediately at a stage before any significant damage has been done.

The success of the screening relies on a high quality programme which pays scrupulous attention to detail and relies on close co-operation between parents, the professionals who link with parents, particularly public health nurses, the Director and team of the National Newborn Screening Programme who process and disseminate the results, and with paediatric staff who treat these children found to have a problem. Like any area of health care, developments in understanding and new techniques extend what is possible.

This review was commissioned by the C.E.O.'s of the Health Boards and the Chief Medical Officer of the Department of Health to ensure that the excellent work undertaken in Ireland continues to keep pace with the best international practice.

At a time when significant changes are occurring in health service structures it will be important to both maintain the integrity of the screening programme and to modernise it in line with the aim of the health service reform.

I would like to thank the review group for their dedication and hard work and for coming forth with recommendations, which will serve to improve the health and well-being of children in the years to come.

Dr. Sean Denyer
Director
HeBE Programme of Action for Children

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# Membership of National Newborn Screening Programme Working Group

Dr. Phil Jennings (Chairperson),
*Specialist in Public Health Medicine, Midland Health Board*

Ms. Liz Doyle,
*Assistant Director Public Health Nursing, East Coast Area Health Board*

Ms. Mary Duff,
*Director of Nursing, Our Lady's of Lourdes Hospital, Drogheda, Co. Louth*

Mr. Peter Finnegan,
*Regional Manager, South Eastern Health Board*

Dr. John Gleeson,
*Consultant Pediatrician, Sligo General Hospital*

Dr. Philip Mayne,
*Director, National Newborn Screening Laboratory, The Children's University Hospital, Temple Street, Dublin*

Ms. Maria Molloy,
*Director Public Health Nursing, Mid-western Health Board*

Dr. Eileen Naughten,
*Consultant Pediatrician with special interest in inherited metabolic disorders, The Children's University Hospital, Temple Street, Dublin*

Dr. Ailis Ni Riain,
*General Practitioner, Irish College of General Practitioners*

Dr. Ailis Quinlan,
*Assistant Director, Best Health for Children*

Dr. Lelia Thornton,
*Specialist in Public Health Medicine, ERHA (To July 2002)*

Ms. Regina Walsh,
*Project Officer, Best Health for Children*

# Acknowledgments

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Fr. Michael G Begley,
*Director of Refugee Research, Spiritan Asylum Services Ireland, Dublin.*

Dr. Harold Brenner,
*Specialist in Public Health Medicine, Waterford.*

Ms. Catherine Butler,
*Director, Irish Refugee Council, Dublin.*

Dr. Elizabeth Cullen,
*Area Medical Officer, NAHB, Health Centre, Parnell West Hotel, Dublin.*

Ms. Majella Darcy,
*Development and Education Officer, Comhalmh, Dublin.*

Mr. Frank Edwards,
*Policy Unit Health Services, Reception and Integration Agency, Department of Justice, Dublin*

Mr. Jean Pierre Eyanga,
*Project Officer, Integrating Ireland, Dublin*

Dr. Margaret Fitzgerald,
*Chief Specialist Public Health, Food Safety Authority of Ireland, Dublin*

Dr. Marita Glacken,
*Specialist in Public Health Medicine, Western Health Board*

Dr. Tessa Greally,
*Specialist in Public Health Medicine, Mid Western Health Board*

Ms. Margaret Hanihoe,
*Community Midwife, Domino Project, National Maternity Hospital, Dublin*

Ms Pamela Howard,
*Policy Unit, Health Services, Reception and Integration Agency, Department of Justice, Dublin*

Ms. Juliette O Imiruaye,
*Midwife, Comhlamh, Dublin*

Ms. Celia Keenaghan,
*Researcher, Best Health for Children, Sligo*

Sr. Breege Keenan,
*Social Worker, The Vincentian Refugee Centre, Dublin*

Dr. Patricia Kennedy,
*Department of Social Policy and Social Work, University College Dublin*

Mr. Frank Lennon,
*Superintendent Welfare Officer, Northern Area Health Board*

Ms. Eileen McGlynn,
*Co-Ordinator, Doras Luimni, Limerick*

Mr. Frank Mills,
*Superintendent Community Welfare Officer, Northern Area Health Board*

Mr. Patrick Mulhair,
*Medical Scientist, National Newborn Screening Laboratory,*
*The Children's University Hospital, Temple Street, Dublin*

Dr. Catherine Murphy,
*Senior Area Medical Officer, Southern Health Board*

Ms. Frances Neilan,
*Child Health Development Officer, Western Health Board*

Ms. Mary O'Connell,
*Public Health Nurse, Northern Area Health Board*

Dr. Freda O'Neill,
*Specialist in Public Health Medicine, ERHA*

Ms. Margaret O'Riordan,
*Midwife, Moycullen, Co. Galway*

Ms. Bernie Price,
*Social Worker, The Children's University Hospital, Temple Street, Dublin*

Primary Health Care Project,
*Pavee Point Travellers Centre, Dublin*

Ms. Geraldine Roche,
*Chief Medical Scientist, National Newborn Screening Laboratory,*
*The Children's University Hospital, Temple Street, Dublin*

Ms. Louise Solan,
*Acting Director Public Health Nursing, Western Health Board*

Ms. Mary Syron,
*Manager, Traveller Health Unit, Western Health Board*

Mr. Sam Walklett,
*Supervisor, Roma Support Group, Pavee Point Travellers Centre, Dublin*

Ms. Eileen Weir,
*Director Public Health Nursing, East Coast Area Health Board*

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CHAPTER 1

# Introduction

## Background

### Metabolic Screening of the Newborn

In 1966 The Children's University Hospital, Temple Street under the guidance of Dr. Seamus Calahane, (Consultant Pathologist) established a National Newborn Screening Programme for Phenylketonuria. The programme was funded by the Department of Health and Children and the Eastern Health Board. It was the first national programme of this nature in the world. Phenylketonuria is common in the Irish population with an incidence of 1:4,500. Since 1966, a number of other conditions have been added to the screening programme:

| | |
|---|---|
| Homocystinuria (1971) | Irish incidence 1:68,000 |
| Classical Galactosaemia (1972) | Irish incidence 1:19,500 |
| Maple Syrup Urine disease (1972) | Irish incidence 1:125,000 |
| Congenital Hypothyroidism (1979) | Irish incidence 1:3,500 |

### Principles of Screening

In 1999, the National Screening Committee in the UK up-dated the international criteria for appraising the viability, effectiveness and appropriateness of a screening programme taking into account a more rigorous standard of evidence[1]. All conditions, included in the National Newborn Screening Programme (NNSP), fulfil, in part or in full, the internationally established criteria for screening;

- Evidence from high quality randomised controlled trials that the screening programme is effective in reducing mortality or morbidity.
- Evidence that the complete screening programme is clinically, socially and ethically acceptable to health professionals and the public.
- Benefit from the screening programme should outweigh the physical and psychological harm
- Opportunity cost of the screening programme should be economically balanced in relation to expenditure on medical care as a whole.
- Plan for managing and monitoring the screening programme and an agreed set of quality assurance standards.
- Adequate staffing and facilities for testing, diagnosis, treatment and for programme management should be made available prior to the commencement of the screening programme
- All other options for managing the condition should have been considered to ensure that no more cost effective intervention could be introduced or current interventions increased within the resources available.
- Evidence-based information, explaining the consequences of testing, investigation and treatment, should be made available to potential participants to assist them in making an informed choice.

## Metabolic Disorders Working Group

In 1990, the Minister for Health appointed a Metabolic Disorders Working Group to review the screening programme. The objective of the Working Group was to draw up procedures and practices to ensure universal screening for metabolic disorders and to facilitate a co-ordinated approach by all Health Boards. Health Boards were asked to outline their practices and procedures in respect of hospital births and domiciliary births in their areas. Based on their findings the working group concluded that there was room for improvement regarding screening procedures. The group reported in 1993[2] and made recommendations regarding:

- Responsibility for ensuring that all newborn infants were screened for metabolic disorders
- Practices and procedures to facilitate this
- Development of a co-ordinated approach by all Health Boards.

Work has progressed on these recommendations specific to screening for metabolic disorders. Subsequent to 1993 representation was made by the Director of the National Newborn Screening Laboratory (NNSL) to the Department of Health and Children highlighting concerns regarding certain aspects of the programme particularly in relation to overall responsibility for the programme. Representation was also made to improve the methodology of the screening programme to the Department of Health and Children, and later to the ERHA.

The publication of *A Practical Guide to Newborn Screening in Ireland*[3] first published in 1999 and up-dated in 2001 included recommendations from the Metabolic Disorders working group around practices and procedures of sample collection.

Recent developments are likely to impact on the NNSP, namely funding has been secured from the Department of Health and Children to run a pilot screening programme for Congenital Toxoplasmosis. Furthermore, a Cystic Fibrosis Screening Working Group has been established at the request of the Chief Medical Officer, Department of Health and Children. Its brief is to identify the resources required to support a high quality newborn Cystic Fibrosis screening programme.

## Best Health for Children Report

The report *Best Health for Children - Developing a Partnership with Parents*[4] which had been commissioned by the Chief Executive Officers of all the Health Boards in the Republic of Ireland was published in 1999. It made many recommendations regarding changes in delivery of the child health services.

The report contains a significant number of detailed recommendations around many issues including the NNSP. The recommendations regarding the latter are:

"Responsibility for co-ordinating the NNSP should be assigned to one body.
Nationally agreed protocols for screening should be drawn up.
An audit of the screening programme should be performed addressing in particular the completeness of cover, and the timeliness of testing and reporting."

## National Newborn Screening Programme Working Group

Best Health for Children (BHFC) was established by the Conjoint group of Chief Executive Officers of all the Health Boards in the Republic of Ireland in 1998 to facilitate implementation of the report. In 2001, BHFC was requested to carry out a review of the NNSP. In order to further this, a working group, comprised of key personnel, reflecting geographical and professional representation was established to oversee this review. A representative from the Department of Health and Children was sought to sit on the Working Group. However since no one individual in the Department of Health and Children was considered to have responsibility for the NNSP, this request could not be fulfiled.

## Terms of Reference

**The terms of reference for the National Newborn Screening Programme Review Group were to:**

**1. Audit current practice throughout the country, with regard to;**

a) Coverage
b) Timeliness of testing and reporting
c) Quality assurance
d) Consumer satisfaction

**2. Development of an overall management structure with regard to;**

a) Responsibility and accountability for screening
b) Development of nationally agreed protocols for screening
c) Set standards for quality assurance and determine performance indicators
d) Identification of resources required to support a high quality system

Two particular categories were deemed by the working group to merit special attention;

1) Irish Travellers
2) Refugee/asylum seekers, as there are issues pertinent to these groups which have implications for the success for the NNSP.

CHAPTER 2

# The Process

The process of newborn metabolic screening is complex and involves many steps.
In summary;

1) Capillary blood sample is taken by heel-prick, when the infant is aged 72-120 hours and collected onto a Newborn Screening Card (NSC).
2) The sample is transported to the National Newborn Screening Laboratory (NNSL).
3) Testing for conditions being screened is carried out at the NNSL.
4) Results are communicated to the maternity unit in a hospital and to a community care area or to a domiciliary midwife.
5) Action taken is dependent on results.

## Timeframe

The Report of the Metabolic Disorders Working Group (1993)[2] recommended a timeframe for sample collection i.e. "after 72 hours and before 120 hours from birth".
The reason for the specific timeframe is that some of the conditions, for example Galactosaemia and Maple Syrup Urine Disease, benefit from the earliest detection possible. Early detection may save the child's life and protect its intellect. If the sample is collected too late after birth some infants may present clinically before the results of the test are available. This is of particular relevance to the Traveller population as the incidence of Galactosaemia is 1 in 450 births in comparison with an incidence of 1 in 36,000 births in the non Traveller population.

Such a specific timeframe is not suitable for all conditions, e.g. the optimum time to screen for Homocystinuria is between day 7 and day 10. Evidence from audits of the National Newborn Screening Programme (NNSP) and of European screening programmes suggests that screening programmes will not detect approximately one in five cases of Homocystinuria. Individuals who are not detected by the programme may present in later life with dislocation of lenses, osteoporosis and tall stature. Severe mental handicap can also result. These cases may not be detected by a newborn screening programme in the case of;

- breast fed infants as there may be an inadequate intake of methionine in the feed to enable detection.
- infants who have a milder vitamin B6 responsive form of the disorder.

The process for metabolic screening is dependent on how responsibility for screening is delegated. The Report of the Metabolic Disorders Working Group (1993)[2] set out recommendations around responsibility stating that ultimate responsibility for ensuring that all infants are screened rests with the Health Board. Hospitals are responsible for ensuring that all infants in hospital are screened for metabolic disorders. If the baby is discharged before testing can take place, the hospital continues to be responsible for ensuring that the child is screened by appointment at the hospital or that responsibility is delegated to the Director of Public Health Nursing to perform the test in the community. If the test is carried out in the community, the case becomes the responsibility of the Director of Public Health Nursing. In the event of a home birth, the midwife and the General Practitioner attending at the birth are responsible for performing the test for all infants under their care.

There has been a significant change in practice with mothers being discharged from maternity units within days if not hours of birth. Thus, arrangements must be made for infants discharged from maternity units before the sample has been taken. Local policy usually dictates that testing may be carried out by the Public Health Nurse in the community, or, the mother may be requested to bring the infant to the maternity unit of birth. A significant proportion will return to the maternity unit. Although, theoretically, testing may be done by General Practitioners, this does not generally occur.

It is evident from the above that three distinct pathways are involved in this system:

1) Sampling is performed on infants born at home.

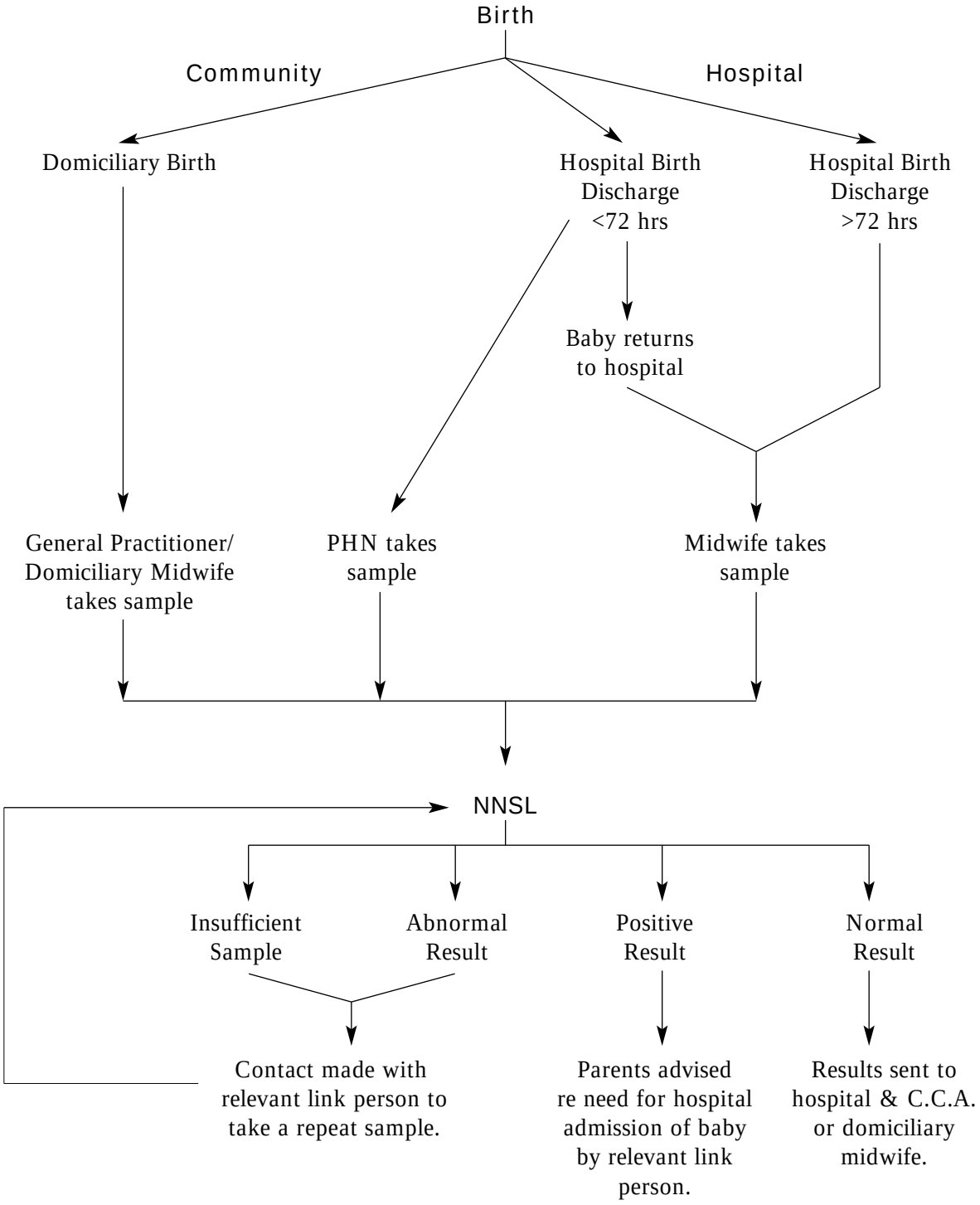
2) Sampling is performed on infants born in maternity units while the infants are in the unit.

3) Sampling is performed on infants born in maternity units following discharge of the infant to the community.

The complexity of the system, is illustrated in the flow-chart on page 12.

## Flow Chart for Metabolic Screening of the Newborn in the Republic of Ireland

(This is a general outline. Modification may be required depending on the individual condition)



CHAPTER 3

# Methodology

A combination of quantitative and qualitative research was used to inform this report.

## Quantitative Research

The Hazard Analysis Critical Control Point (HACCP)[5] approach is commonly applied to food safety. The working group considered that it would be of benefit to apply this system to the National Newborn Screening Programme (NNSP). Thus, the critical points in the system can be identified, as can the controls necessary to guarantee safe practice and 100% coverage. The Hazards Analysis Critical Control Point Approach (see appendix 1) accurately describes the entire process of metabolic screening to ensure that all potential hazards may be identified. This approach was used to inform the development of a questionnaire with the objective of examining the entire process.

## Questionnaire Development

The questionnaire addressed the following areas:

- Responsibility
- Protocol
- Liaison
- Weekend Testing
- Recording
- Transport of samples
- Reporting
- Consent
- Refusal
- Parental Information
- Design of Newborn Screening Card (NSC)
- Training

The number of open-ended questions was kept to a minimum.
The questionnaire was piloted on a sample of the relevant study population to ensure validity, reliability and consistency.

## Data Sources

The main sources of data for this survey were from the mailing lists of the (National Newborn Screening Laboratory) NNSL and the domiciliary Midwives Association respectively.

## Study Population

74 questionnaires were sent to the following groups and response rates are as shown below;

| *Respondents* | | *Response Rate* |
|---|---|---|
| Masters of the Dublin Maternity Hospitals | (3) | 100% |
| Directors of Nursing | (19) | 100% |
| Directors of Public Health Nursing | (32) | 100% |
| Practicing Domiciliary midwives | (11) | 68% |
| Total | (65) | (93%) |

The overall response rate was 93%.

## Data Analysis

The data was analysed using EPI Info 6.

## Qualitative Research

The qualitative research took many forms;

**A) Site visit by the working group to the NNSL**

In the Republic of Ireland, newborn metabolic screening is carried out only in the NNSL. To fully comprehend the complexity of the screening process, the working group visited the NNSL and observed the steps involved in the laboratory testing process for national newborn metabolic screening.

- Arrival of Newborn Screening Cards (NSCs)
- Preparation of cards for testing
- Analysis of sample
- Recording results
- Liaison
- Dissemination of results

**B) Interviews with the Medical Director of the laboratory**

A number of interviews were carried out with the Medical Director of the NNSL. He identified issues which needed to be considered:

- Overall management structure of the Screening Programme with the establishment of a 'Board' responsible to the Department of Health and Children (or HeBE)
- Funding of the Newborn Screening Laboratory
- Technology to support a quality assured service
- Computerisation with links to maternity units, community care areas, birth registers and Health Boards. This would also facilitate audit.
- Training of those involved in specimen collection
- Redesign of NSCs
- Storage of NSCs
- Consent and refusal

**C) Interviews with representatives of Traveller and Refugee support organisations**
Interviews and meetings were held with support organisations and interest groups to explore issues pertinent to ethnicity and newborn metabolic screening. These included:

- Communication between service users and service providers
- Provision of health promotion information
- Sensitivity of health service staff to cultural differences.

**D) Interviews with mothers of recently born babies**
Interviews were held with a small group of mothers, who had recently had babies to establish;

- Their level of information concerning the "heel prick" test.
- Their views on the process, including consent and refusal.
- Any recommendations for improvement.

CHAPTER 4

# The Maternity Units

Questionnaires were sent to the Masters of the three major Dublin maternity hospitals and the Directors of Nursing in nineteen maternity units in the Republic of Ireland. A 100% response was received from this group. Analysis of the questionnaire yielded the following information.

## 1. Responsibility

**Overall responsibility**
*The Report of the Metabolic Disorders Working Group (1993)*[2] stated "The Group recommends that hospitals should be responsible for ensuring that infants born in hospital are screened for metabolic disorders". The report does not however designate any one person in each hospital as having ultimate responsibility for metabolic screening.

The Masters of the three large Dublin maternity hospitals have responsibility for the metabolic screening in their respective hospitals. In all other hospitals, however a wide variety of personnel are nominated as being responsible for screening for metabolic disorders.

**Responsibility for Metabolic Screening at Ward Level**
At ward level responsibility for metabolic screening is generally held by a variety of nurse management and midwives. In a number of hospitals this responsibility is co-shared.

## 2. Protocol
Application of the Hazard Analysis Critical Control Point (HACCP) approach to the newborn metabolic screening procedure identifies many critical points. To standardise the process of newborn screening testing the National Newborn Screening Laboratory (NNSL) developed *A Practical Guide to Newborn Screening in Ireland (2001)*[3] The majority of hospitals follow these guidelines. Other hospitals have developed their own protocol in addition to or instead of the NNSL Guidelines.

## 3. Weekend Testing
The majority of hospital respondents consider that weekend testing should be available in the community. It must be recognised that weekend newborn screening testing takes place in no other country in Europe. Convenience for parents, the practice of early discharge, and the reduction of workload for hospital weekend staff were cited as the main reasons for its availability.

*A Practical Guide to Newborn Screening in Ireland (2001)*[3] specifies that the sample for Newborn screening testing should be taken when the infant is aged between 72 and 120 hours and should reach the laboratory as soon as possible after collection. In practice, where weekend testing in the community is not available, hospitals advise parents to bring the infant to the out-patients department of the maternity unit for newborn screening testing during a weekend period. Alternatively parents are advised that the PHN will carry out testing on the following Monday.

A small number of hospital respondents also raised the issues of staff shortages in the community care areas, the absence of weekend transport to deliver tests when taken, and the fact that the NNSL does not provide a routine weekend service. They therefore queried the need for any weekend testing service.

**Transport of Samples**
The transport of samples at the weekend can pose problems for some hospitals. A variety of forms of transport are used in dispatching samples to the NNSL to ensure they arrive as soon as is practicable after collection. These include registered or regular post, courier, taxi and train. When using post, hospitals may place more than one sample in an envelope.

## 4. Liaison

Infants born in any one maternity unit may be discharged to a large number of different community care areas and in some situations to different health board regions. Maternity units are required under the Notification of Births Act 1905 and 1915 to notify community care areas within 36 hours of a birth. A study in the Western health board has shown this occurred in only 65% of births, emphasising the need for an effective liaison system[6]. Regarding newborn screening testing, there are three scenarios in which liaison is required. These are:
(a) Discharge of an infant who has not yet been tested into the community
(b) Linking with the NNSL regarding results
(c) Reporting results to appropriate parties.

Hospital liaison procedures are developed around the workings of the individual hospitals rather than following a national standard. In a number of hospitals the same person is responsible for all liaison regarding newborn screening testing. However this is not always the case. Variation also exists amongst the hospitals regarding the personnel involved in liaison.

**Frequency of Liaison**
Liaison between the hospitals and community care areas is well established, with many hospitals reporting that liaison takes place five days a week.

**Liaison Regarding Newborn Screening testing Following Discharge**
*The Report of the Metabolic Disorders Working Group (1993)*[2] recommends that a ward sister should be nominated as the person with responsibility for notification to the appropriate Director of Public Health Nursing. Sisters in charge of wards and midwives tend to be responsible for notifying community care areas if an infant is discharged prior to the newborn screening testing.

**Liaison with National Newborn Screening Laboratory**
Twice a week, normal results are posted from the NNSL to a liaison person in the relevant maternity unit. The designated liaison person responsible for receiving these results tends to be a nurse manager. The same individual generally contacts the NNSL regarding missing results by phone or by fax.

**Reporting Results**
A significant number of hospitals have a named liaison person with designated responsibility for reporting results. This role tends to be assigned to nurse managers. The Liaison person ensures that test results are reported directly to Directors of Public Health Nursing, General Practitioners and parents, as deemed appropriate. Parents are more likely to be contacted directly by the hospital in the case of a positive result.

## 5. Recording

Recording of Sample Taking

It is common practice in all hospitals to record that the sample has been taken in the hospital. Responsibility for recording usually rests with the person who performs the test. On discharge, all hospitals record if a newborn screening test has yet to be administered. This information is recorded in a variety of locations. These include the child's record, discharge sheet, birth notification form and in keeping with *A Practical Guide to Newborn Screening in Ireland (2001)*[3] a separate screening register.

**Recording of Dispatch of Sample**

Practice varies from unit to unit regarding recording dispatch of the Newborn Screening Card (NSC) from the maternity unit including;

- Keeping a record of dispatch of samples
- Place of storage of receipt of registered post

**Recording Receipt of Results**

Hospitals vary with regard to the type of results they record with more hospitals recording positive results than negative results. In the case of repeat, query positive or positive results, the liaison person in the hospital is contacted. A staff member is nominated to check that results have been recorded in the majority of hospitals. This check may be carried out on a daily, weekly or monthly basis, or performed on date of receipt of results.

Results of samples taken in the community are also sent by the NNSL to the relevant maternity unit.

## 6. Consent

All respondents state that parents are informed about the purpose and the timing of newborn screening testing. However, there is some evidence to suggest that not all individuals involved in the sampling process are fully informed of the procedures at the time of sampling and not all mothers are informed that the newborn screening sample had been taken. Although the NNSL guidelines state that the mother should be present when the sample is being taken, it would appear that this does not always occur. Being present at the time of sample taking allows the mother to hold the infant when the sample is being taken, ask questions if she wishes, act as a witness to the sample taking and sign a consent form if one is being used by the hospital. However it is the mother's decision whether or not she is present at sample taking.

**Form of Consent**

Consent may be implied or specific. Implied consent is the accepted practice in the majority of hospitals. Furthermore, it is rare for hospitals to obtain written consent. Written consent is considered good practice for screening[7]. Consent may not always be sought. Parents are generally asked to consent to newborn screening testing either prior to screening or at the time of sample collection.

**Refusal**

Written refusal forms, which must be signed by a parent, are rare. Hospitals would generally notify the NNSL in the case of parental refusal for newborn screening testing. The Supreme Court recently affirmed the right of parents to refuse newborn screening testing for their infant[8]. The management of the maternity unit has no option other than to comply with the parent(s)' wishes to decline the screening test.

## 7. Parental Information

Provision of meaningful information to parents is empowering and this has been acknowledged in a number of recent reports[4,9]. A recent report examining health literacy in Ireland recommended the use of simple language in health information leaflets[10]. Health promotion leaflets regarding the NNSP published by The Department of Health and Children are widely available. Hospital respondents considered it would be helpful if additional information was provided to enable them to support and inform parents in this regard. Information needs of ethnic minorities and refugees require particular attention.

### Information Provision

#### a) Information re newborn screening testing

Parents begin receiving information regarding the newborn screening test at their antenatal class. However information is most frequently provided during the immediate post-partum period and at the time of sample collection.

Verbal advice and information leaflets are the most common methods of presenting information to parents.

#### b) Information re Timing and Site of newborn screening testing

Several hospitals give parents a choice of returning to the hospital or having the sample taken in the community. If the sample is to be taken in the community, it is vital that when the NSC is filled out by hospital personnel the correct information is given on the card. This information must be checked by the individual taking the sample and correctly dated. Incorrect information or lack of information on the NSC can lead to time delays in returning results which may have serious consequences if the NNSL is seeking a repeat sample. When a sample is being taken in the community, it is normal practice for hospitals to explain the importance of timing of sample taking to parents.

#### c) Information Provision for Travellers

A number of hospitals provide specific information appropriate to the needs of Travellers. A Practical Guide to Newborn Screening in Ireland (2001)[3] recommends that the Beutler test should be routinely offered to all Traveller infants and siblings of known cases of Galactosaemia, owing to the high incidence of the condition amongst these groups. The Beutler Test is a test for Galactosaemia, testing for the red cell GALT enzyme activity. This test should be carried out prior to ingestion of breast milk or cow's milk, on Day 1 and prior to any blood transfusion. The results of the Beutler test are known within 4 to 6 hours of testing in the NNSL. In the event of a positive test result, the hospital is telephoned directly. However resource constraints prevent the NNSL from telephoning negative results to hospitals and these are sent by post to the hospital. Up-dated information leaflets are required to provide information on the Beutler test.

#### d) Information Provision for Ethnic Minorities and Refugees

Maternity units are attempting to cater for the specific needs of refugees and asylum seekers. Translation services are available in certain hospitals however research has indicated that consumer satisfaction with this service has been low[11].

A major concern for hospitals is the need for provision of culturally appropriate information in a variety of languages, in leaflet form, for ethnic minorities and for refugees. At the time of this survey, one hospital had leaflets available in Russian, Romanian, French, Czech, Chinese, Albanian, Serb/Croat, Polish, Portuguese, Spanish and Arabic. However, a number of hospitals indicated that they were independently developing leaflets in a number of languages.

**Information leaflets**

The majority of respondents consider there is a need to up-date existing Department of Health and Children information leaflets. In general, more information should be provided regarding:

a) the metabolic disorders tested for
b) rationale for testing
c) information regarding repeat testing
d) the implication of a positive result.

## 8. Newborn Screening Cards

Respondents were invited to comment on the current NSCs. Suggestions included, production of a larger card with more space for writing, smaller circles for blood sampling on the card and examination of the need for the entire card to be made of absorbent material. Some respondents stated that the mother's details rather than the infants should be entered on the card. Several commented on the need for a unique personal identifier number. Addressograph labels are currently in use in a number of hospitals.

CHAPTER 5

# The Community

The questionnaire was sent to thirty-two Directors of Public Health Nursing and a response rate of 100% was achieved. Data analysis revealed the following:

## 1. Responsibility

*The Report of Metabolic Disorders Working Group (1993)*[2] states that the Health Boards have ultimate responsibility to ensure that the infants are screened. The Director of Public Health Nursing has responsibility for all those screened in the community if informed by the maternity unit.

Several respondents named the CEO as having overall responsibility at Health Board level for newborn screening testing. The majority of respondents named the Director of Public Health Nursing as having responsibility at community care area level. The Assistant Director of Public Health Nursing was most commonly identified as having responsibility for co-ordinating the programme. However a wide range of personnel were identified as having responsibility at all levels.

## 2. Protocol

*A Practical Guide to Newborn Screening in Ireland (2001)*[3] is used by a large number of community care areas. Some community care areas have developed their own written protocol to be used either in conjunction with or instead of this guide.

Difficulties that community care areas encounter in adhering to protocol include the following:
- Location of migrant groups
- Failure to receive notification that test is to be collected
- Insufficient information on Newborn Screening Card (NSC)
- Insufficient time between sample taking and postage to allow complete drying of sample
- Absence of weekend postal service leading to time delays in dispatch of samples
- Cross border births and differing protocols
- Informing parents regarding need for repeat testing and test in general.

## 3. Weekend Testing

Several community care areas provide a weekend newborn screening testing service. At weekends, in community care areas where community testing is not available, it is customary for mothers to be advised to return to the maternity hospital for testing. Some feel that there is a need for weekend testing in the community. Distance from maternity hospitals, transport difficulties for parents, and testing within the recommended time frame are the reasons given for provision of community weekend testing. However barriers to weekend testing included:

a) Manpower shortages
b) Absence of week-end postal service
c) Absence of routine week-end laboratory testing
d) Need to enhance the hospital-community liaison service.

**Transport of Samples**
Community care areas tend to use a variety of transport options when sending samples to the National Newborn Screening Laboratory (NNSL) from Monday to Friday, at weekends or during Bank Holidays. Registered post is the most commonly used mode of dispatch. Hospital transport, courier and train are also used. It is sometimes possible to use registered post/regular post at the weekends but only in areas where the local post office is open on Saturday.

## 4. Liaison

To ensure that every child is tested within the recommended time frame, the appropriate information must be forwarded to the community by the hospital in a timely manner. The majority of community care areas regard newborn screening test requests from hospitals as timely. Liaison takes place 5 days a week in most community care areas.

**Hospital - Community Liaison**
Due to the increasing number of early discharges, more newborn screening tests are being administered in the community, and thus, effective liaison is of paramount importance. Consequently a community care area may have a designated hospital- community liaison person. Public Health Nurses are commonly assigned to this role. This role may also be shared amongst a variety of personnel in community care areas.

**Liaison with the National Newborn Screening Laboratory**
The NNSL sends a copy of all results to an identified individual within a Community Care Area, irrespective of whether a test is performed in a hospital or by a Public Health Nurse in a community care area. However not all nurses who take a sample in the community receive feedback that the sample has been received by the laboratory or the test result. Almost all community care areas have nominated a person to receive newborn screening test results. This role is commonly assigned to the Assistant Directors of Public Health Nursing.

**Communication of Results**
The majority of community care areas have a named liaison person with designated responsibility for reporting results.

Notifying parents of results, especially a positive result or a repeat result requiring another test, is a delicate task, which must be done in a professional manner. It is acknowledged that the more information parents receive around the nature of their child's possible condition, the less anxiety they will feel during this stressful time. The NNSL has developed a procedure for reporting results including the information that will be required by the contact person to answer correctly any queries parents may have. This procedure is relayed from the NNSL, to an initial contact person, generally situated in the hospital. This contact person may then ask the Public Health Nurse to locate and inform the parents of the result in the community. Owing to the relatively low incidence of the conditions being screened for it is possible that the Public Health Nurse may not be entirely familiar with the condition. Thus the role of the PHN is to support the parents to access the hospital at this stage to get detailed information.

## 5. Records

All community care areas have a recording system in place for newborn screening testing. The majority of community care areas record a hospital's request for a newborn screening test and also record that the newborn screening test has been collected.

*A Practical Guide to Newborn Screening in Ireland (2001)*[3] recommends that Directors of Public Health Nursing should keep a register of requests from hospitals. In the majority of community care areas, these records are kept in dedicated screening registers. Responsibility for recording this information varies but tends to be performed by Public Health Nurse Managers or Liaison Public Health Nurses. Community care areas generally have a mechanism in place to ensure that this information is recorded.

**Recording Dispatch of Sample**

As recommended in *A Practical Guide to Newborn Screening in Ireland (2001)*[3] all community care areas record individual samples sent to the NNSL but again in a wide variety of locations. All keep receipts of registered post if used. In general, only one certificate of postage is obtained if more than one test sample is enclosed in an envelope. As a result of this practice, it is not possible to confirm that individual samples have been included in batches sent to the NNSL.

**Recording Receipt of Results**

All but one community care area keeps a record of all test results. The screening register is most commonly used. Community care areas tend to have a nominated person responsible for checking that test results have been recorded. Weekly checks of results are performed in most community care areas.

## 6. Consent

Verbal consent is obtained in the majority of instances, with few community care areas obtaining written consent. Information is most frequently provided at the time of sample collection.

**Refusal**

*A Practical Guide to Newborn Screening in Ireland (2001)*[3] states that if parents refuse to allow their infant to be screened they should be requested to indicate their refusal in writing and that a copy should be forwarded to the NNSL. Community care areas that have refusal forms are in the minority. In the event of a refusal, it is the policy in most community care areas to contact the NNSL.

## 7. Parental Information

*The Report of the Metabolic Disorders Working Group (1993)*[2] recommended that an information leaflet for parents be published in order to increase the level of awareness of the importance of screening and the consequences of delay in detection of the condition being screened. These leaflets are available through the Health Promotion Unit, Department of Health and Children. However it is recognised that these leaflets require up dating.

To date, no community care area has formally evaluated parental satisfaction. One community care area is currently developing a policy on evaluating parent satisfaction of newborn screening testing.

**Information re newborn screening test**

Almost all community care areas provide information concerning newborn screening testing to parents at the time of sample collection, often through information leaflets and verbal advice.

**Information for Travellers**
Half of the community care areas cater for the specific information needs of Travellers.

**Information for Ethnic Minorities**
A little over half of the community care areas provide translation services for refugees while less than half cater for the specific information needs of refugees.

**Information Leaflets**
The majority of community care area respondents feel that existing information material should be up dated. Three areas identified as needing attention were the need for:
• Information regarding the test procedure
• Health promotion materials in simple and appropriate language
• Information to be available in different languages and be culturally appropriate.

## 8. Training and Equipment

*The Report of Metabolic Disorders Working Group (1993)*[2] recommended an educational programme for professionals involved in specimen collection. Under legislation, Public Health Nurses are required to visit mothers within 48 hours of discharge from hospital. Owing to manpower problems and at holiday times, registered nurses may replace Public Health Nurses on these visits and thus take the samples in some community care areas. This should not be a cause of concern once the registered nurse receives the appropriate training regarding the metabolic screening process.

**Current Situation**
Most community care areas provide induction training than provide refresher training to Public Health Nurses. Local maternity hospitals and the NNSL play a part in the training process. To date the NNSL has provided training in Galway, Roscommon, Tralee, Sligo, Limerick, Cork, Kilkenny and the three major Dublin maternity hospitals.

**Future**
Sample takers would like to receive additional information on every aspect of the National Newborn Screening Programme (NNSP). Community care staff are interested in training and information in relation to test technique, test results and the delivery of results to parents. Public Health Nurses want regular up-dating and in-service training on the NNSP. It has been suggested that Directors of Public Health Nursing are the most appropriate persons to organise future training as they are aware of the particular training needs of personnel in the community care area.

*A Practical Guide to Newborn Screening in Ireland (2001)*[3] provides a list of equipment needed to take a sample for newborn screening testing. This is available to all sample takers in the community, but is not available in pack form. In a submission to the working group (2001), Public Health Nurses in the ERHA suggested that the NNSP should undertake to develop a sample "taking pack". The NNSL is in the process of developing a standard for the contents of such a pack. Community care areas can then use this to inform the development of their own NNSP sampling pack for Public Health Nurses in their region.

## 9. Newborn Screening Card

Respondents from community care areas made a number of suggestions regarding changes to the NSC. These included;

- Increasing the size of card to allow more space to record patient details
- Reducing the size of the circles for blood sample
- Incorporation of space for written consent
- Pre-printed details
- Production of a duplicate form.

Others queried the possibility of using different paper, which would allow the sample to dry more quickly, and some "indicator" to confirm that an adequate sample had been obtained.

CHAPTER 6

# Domiciliary Midwives

In 1999, two hundred and sixty two domiciliary births were recorded. Although no more recent data are available, it would appear that there may be a trend for an increasing number of mothers to opt for home births.

The questionnaire was sent to sixteen domiciliary midwives, known to be in active practice. A response rate of 68% (11) was achieved.

## 1. Protocol

The majority of domiciliary midwives, who responded to the questionnaire, were aware of the National Newborn Screening Laboratory (NNSL) protocol for newborn screening sampling. Some complained of the difficulty in adhering to the protocol resulting in delays in sample collection. Reasons for delays cited were mothers breast feeding their infants, allowing both parents time to make an informed choice regarding newborn screening, and absence of weekend postal service.

## 2. Weekend testing

Some domiciliary midwives feel there is a need for a weekend service was to ensure compliance with the time frame for sampling as set out in the NNSL protocol. Registered post is commonly used by domiciliary midwives to dispatch samples to the NNSL over weekends or during Bank Holiday periods.

## 3. Liaison

### Sampling

The channel of communication between a domiciliary midwife and the NNSL is direct. Following a domiciliary birth, the midwife generally takes the sample and then forwards it to the NNSL. In some cases, a parent may be requested to post the sample. Although there is no statutory requirement to notify or involve any other party, respondents state that they routinely notify the relevant community care area that newborn screening testing has been taken. This information allows community care areas to check that a sample has been taken if a query arises.

### Reporting Results

The NNSL guidelines clearly state that results from normal samples should be directed to the initial sample taker when the appropriate and relevant information has been put on the NSC. However domiciliary midwives state they do not receive normal test results.

In the event of a repeat result, the NNSL guidelines suggest contacting the liaison person at the Maternity unit or the Director of Public Health Nursing. Acting on a request from the NNSL for a repeat sample the Director of Public Health Nursing may contact the domiciliary midwife responsible for the delivery or the relevant PHN. Practice appears to vary from area to area. The NNSL confirmed that the Director of Public Health Nursing rather than the domiciliary midwife may be contacted if the original contact details for the domiciliary midwife on the NSC are incomplete.

## 4. Recording

The NNSL guidelines make recommendations regarding recording arrangements in both hospital and community care setting but make no specific reference to record keeping by Domiciliary midwives. However, all domiciliary midwives keep a record of samples sent to the NNSL.

## 5. Consent and Refusal

All respondents report that parents are informed about the purpose and timing of the newborn screening test, prior to gaining consent. Implied consent is generally obtained by domiciliary midwives at the time of sample collection. Few respondents have access to written consent or refusal forms. Additionally only a small number of domiciliary midwives would inform the NNSL in the advent of a refusal

## 6. Parental Information

Parental information given during the antenatal period or at the time of sample collection is usually given verbally. In some instances satisfaction with newborn screening testing information is evaluated verbally.

**Information provision for Travellers**

The rate of domiciliary births is very low amongst Travellers and no domiciliary midwife caters for the special needs of Travellers.

**Information provision for refugees and asylum seekers**

No domiciliary midwife caters for the special needs of refugees and asylum seekers. Domiciliary midwives reported having no access to translators or information leaflets in different languages.

**Information leaflets**

Suggestions to up-date information leaflets included the provision of information around the purpose of the test, description and incidence of each disorder, and relevance of timing of the test.

## 7. Information, Training and Equipment

Domiciliary midwives may receive induction training in the community prior to carrying out newborn screening testing and up-date training. Domiciliary midwives who do not currently have a copy of *The Practical Guide to Newborn Screening Guidelines (2001)*[3] state they would like information on newborn screening testing and the NNSL. Some domiciliary midwives appear to be unaware that these guidelines are available free of charge from the NNSL.

## 8. Changes to the content or layout of the card

Domiciliary midwives believe that the card has little relevance to a domiciliary birth and have suggested that the card should be amended to incorporate the following; place of birth, individual record of mother's surname and infant's surname and contact details for domiciliary midwife.

CHAPTER 7

# The Laboratory Perspective

## Laboratory Services

The screening, diagnosis and biochemical monitoring of infants, children and adults with inherited metabolic disorders relies on the facilities of at least two related laboratory services, both based at the Children's University Hospital, Temple Street. These include:

**1. The National Newborn Screening Laboratory (NNSL)**

**2. The Tertiary Referral Metabolic Laboratory.**
This laboratory provides a comprehensive service for the life-long monitoring of individuals diagnosed through the newborn-screening programme and for the diagnosis of many other inherited metabolic disorders in infants, children and adults who present with clinical symptoms. In addition samples are referred nationally from infants, children and adults on whom clinicians suspect the possibility of an inherited metabolic disorder.

Staff within these two laboratories work in close association with clinical, nursing and dietetic staff in the National Centre of Inherited Metabolic Disorders also located at the Children's University Hospital and with staff in other hospitals throughout the country.

In the Republic of Ireland, all samples for newborn screening are analysed centrally in a single laboratory, the NNSL based at the Children's University Hospital, Temple Street, Dublin.

The working group visited the NNSL at the invitation of the Medical Director, Dr. Philip Mayne.

## Testing Process at National Newborn Screening Laboratory

Approximately 58,000 specimens (290,000 tests) are processed each year. This number includes samples from approximately 99.9% of infants born in the Republic of Ireland, and repeat samples. A small number of Irish resident parents opt to have their infants born in Northern Ireland and many of these infants are initially screened in the North. The number of specimens received and processed by the NNSL is in accordance with the optimum size of a newborn screening laboratory as discussed by the Directors of UK Screening Laboratories during their Annual General Meeting of 2000.

## Arrival of Newborn Screening Cards

National Newborn Screening Cards (NSCs) arrive at the NNSL during weekdays and on Saturday mornings from:

- Maternity units
- Special care units in paediatric hospitals
- Individual Public Health Nurses from community care areas
- Domiciliary midwives.

Two clerical staff enter infant demographic details from the cards onto a computerised database (MediSense Apex Laboratory Information System). Additional information detailed on the NSC, such as breast feeding information, and antibiotic treatment is not recorded. Issues that typically arise at this stage of the procedure include:

- Mother's addressograph label with mother's Hospital Number (rather than infant's) used on NSCs
- Incorrect or no community care area code
- Provision of an inadequate blood sample
- Omission of the date of sample collection
- Poor handwriting
- Possibility of incorrect linkage of mother's name and baby's name
- Possibility of incorrect linkage of mother's hospital number and baby's hospital number.

## Preparation of Newborn Screening Cards for Testing

To ensure suitability for testing, the four blood spot rings on the NSC should be soaked through from the rear to the front of the card and be completely filled with blood. The blood spots must be dried before they are packaged and dispatched to the NNSL.

On receipt, the NSC is inspected for its suitability for analysis. It is assigned a unique number.

## Analysis of Sample

The assay procedure, based on semiquantitative bacterial inhibition assay (BIA) and first developed by Dr Robert Guthrie New York (USA) in 1958, is used in testing for;

- Phenylketonuria
- Classical Galactosaemia
- Maple Syrup Urine Disease
- Homocystinuria

A 3mm spot for each assay is punched from the "autoclaved" sample. This spot is placed on a previously prepared agar plate, specific for a bacterial inhibition assay. Each tray contains a batch of 56 samples, standards and controls. A number is assigned to each "spot" in order to link it to the infant. This number is used to identify all 'spots' taken from the same sample.

An immunofluorometric assay is used to screen for Congenital Hypothyroidism. Spots for analysis for congenital hypothyroidism are punched manually directly in 96-well microtitre plates.

The BIA procedure however lacks sensitivity and precision particularly at the lower end of the analytical range. Three of the four assays results are interpreted visually against a set of standards. Thus, the interpretation of results is operator subjective. To address this concern, two independent, trained medical scientists read all plates. The procedure is lengthy, as it requires over-night incubation.

## Recording Results

Results are recorded as batch results. Individual test results are not recorded. The batches tested are then disposed of and results are documented in a diary and also entered into the computer against the infant's demographic record.

## Liaison

There is no clinical liaison officer in the NNSL, to liaise with maternity units and community care areas. Liaison is required to request repeat samples, organise re-testing, and tracking and following up on repeat test requests, in addition to arranging admission of cases with positive results. At present, this important but time-consuming function is undertaken by the senior medical scientist and by the Medical Director.

## Results

### a) Normal Results

A list of all normal newborn screening test results is issued approximately twice each week. If sampling has been carried out in a maternity unit, a spreadsheet containing the results of samples taken there is sent by post to the relevant maternity unit, with a copy to the relevant community care area(s). In the case of community sampling, a report is sent to the community care area of origin and a copy to the maternity unit or to the domiciliary midwife. The time interval between receipt of sample and posting of normal results is dependent on the day of arrival of sample at the NNSL and whether any one of the five tests has to be repeated. Thus, the interval can range between two days and five days. A copy of the report is not sent to the General Practitioner.

### b) Requests for Re-tests

Approximately 1,200 re-tests are carried out each year. These are requested because of:

- Equivocal or borderline test result
- Insufficient blood for full analysis
- Unsatisfactory analysis
- Assay failure or poor quality control results
- Difficulty in interpretation owing to contamination or deterioration of the sample
- Infant less than 72 hours of age when blood sample was collected
- Card inadequately dried prior to placement in plastic coated enveloped causing ‘serum rings’.

Requests for repeat samples issued by the NNSL are sent by post due to resource constraints. Faxing is rarely used to request a repeat sample. Faxing requests is seen as unsafe unless there is a dedicated person in the hospital or community care area to check the fax machine regularly for requests.

If a repeat sample is requested because of inadequacy of blood collection, the original sample will still be analysed but no result issued. This is a safety procedure, just in case a grossly abnormal result is obtained and consequently the clinician can be alerted to the possibility of a disorder and asked to review the infant clinically.

Sample takers are requested to indicate on the NSC that a repeat test has been sought by the NNSL. This request is rarely complied with, leading to repeat tests being treated as initial tests. Discrepancies in details regarding mother’s and the baby’s name, a change in the infant’s surname or first name, the date of the sample, and the number of the original card may give rise to administrative queries and lead to delay in return of results.

**c) Query Positive Tests**
Query positive test results suggest that an infant may have one of the conditions being screened for. Supplementary tests are needed to confirm the diagnosis. The response to these cases is immediate and direct. The liaison person in the maternity unit is contacted by telephone and requested to arrange for the child to be brought directly to The Children's University Hospital, Temple Street or to the local Paediatric unit as appropriate. Technical staff or the Medical Director arrange admission to the Children's University Hospital and inform the Metabolic Unit of the detail of the pending admission.

When possible, the Medical Director or the medical staff from the Metabolic Unit will meet with parents on arrival at the hospital, after a further blood sample has been taken. The Medical Director explains to the parents the reason for the admission and then informs the parents of the results when available. Positive cases are admitted to the wards and false positive cases are referred the same day for a clinical examination and possible further follow-up. The procedure outlined varies depending on the condition under consideration.

Issues discussed with the Medical Director
During interviews with the Medical Director, the following issues emerged:

## Management Structure

*The Report of Metabolic Disorders Working Group (1993)*[2] clearly and unambiguously identifies responsibilities of organisations and individuals but does not designate overall responsibility for the entire National Newborn Screening Programme (NNSP). Such a body is required in order to;

- Audit the programme on an annual basis
- To make evidence based decisions regarding inclusion of additional conditions in the screening programme
- To ensure that all research involving test samples is carried out in a fashion that complies with agreed ethical guidelines.

## Funding

**Revenue:** Funding for the NNSL is allocated through the Eastern Regional Health Authority (ERHA) and is included within the Children's University Hospital annual allocation but is not identified and consequently not ring fenced. Appendix 2 details the funding costs for 2003. The NNSL submits an annual strategic plan to hospital management, and this is included within the hospital's submission to the ERHA. The NNSL although providing a national service is thus competing for resources with other services within the hospital itself. This is obviously unsatisfactory for both the hospital and the NNSL.

**Conferences/International management meetings:** Funding is required for the Medical Director and staff to attend international meetings to keep up to date with developments in newborn screening. These meetings include the annual UK Directors meeting, the biannual Euroscreen Meeting (the European Directors screening meeting) and the International Society of Neonatal Screening Conference, in addition to specialist meetings, for example on Toxoplasma and CF. Attendance at these meeting is currently paid for by the Children's University Hospital, the Medical Director's CME allocation and by the Medical Director himself.

**Capital:** Equipment replacement is prioritised both within the Pathology Division and also within the hospital with regard to funding. As a consequence, funding competes for limited resources with other essential equipment such as ICU/theatre ventilators and ICU equipment. There is no hospital or ERHA accepted planned replacement strategy.

## Technology/Methodology

The methodology used within the NNSL has remained essentially unchanged since being introduced, initially in 1966. The bacterial inhibition assays do not conform to Good Laboratory Practice (GLP) nor would they withstand the rigors of Laboratory Accreditation procedures.

Tandem Mass Spectrometry (TMS/MS) is now being employed throughout the world and is the recognised method of choice, providing a rapid, sensitive quantitative method. Up to 20 inherited disorders of amino acid metabolism and of fatty acid oxidation can be screened from a single 3mm blood spot. In excess of 50 screening laboratories worldwide are now using MS/MS as the method of choice. Existing semi-quantitative screening methods are being replaced and additional conditions are being screened, depending on the local incidence.

**TMS/MS test has many advantages:**

- Its sensitivity and specificity are significantly higher than with current methods in use in the NNSL.
- It allows for the screening of up to 20 inherited disorders thus allowing the programme to develop over time. This technique has been validated for three aminoacidopathies currently being screened for in the Republic of Ireland, namely, PKU, MSUD and HCU but is not at present suitable for testing for Classical Galactosaemia or Congenital Hypothyroidism.
- TMS/MS may be used for high risk screening in samples obtained from older infants and children suspected of having a possible inherited metabolic condition. Approximately 15 to 20 samples per week are being sent from Ireland to the U.K. for analysis by MS/MS at a cost of 90EUR per sample, excluding handling and transport cost. The current turn around time from receipt of the sample at the Tertiary Referral Metabolic Laboratory to result averages 10 days, with a range of 5-12 days. This has the potential to be reduced to about 4 hours in urgent situations.

A formal request was made to the Department of Health and Children in 1999 for the implementation of Tandem Mass Spectrometry. However, to date, the NNSL has been unable to access funding. Funds have been raised by the Fund-raising Department of the Children's University Hospital to purchase a TMS/MS for high-risk screening. However, this will not be available to the NNSL but could be used as a back-up instrument in the event of instrument failure if Tandem Mass spectrometry is introduced as part of the Newborn Screening testing process.

## Computerisation

In late 1999, data on Labstar was transferred onto a module on the Pathology MediSense Apex Computer. This Laboratory Information System does not have dedicated screening software and as a consequence, recovery of data is restrictive. Consideration should be given to purchasing Perkin Elmer Health Science dedicated newborn screening software to replace the current system well in advance of the laboratories transfer to the Mater Site and having a dedicated, stand-alone computer system. In addition to its data retrieval and management functions and unlike the current MediSense Apex Computer, this dedicated software is compatible with an optical card reader. The advantage of this is that an optical card reader will input patient demographic data and thereby reduce transcription errors.

"Medibridge", is currently being piloted, to assess the feasibility of establishing a computerised data transfer system between the NNSL and Maternity Units. The pilot project initially involves the Coombe Women's Hospital. "MediBridge" is a software product for the electronic transfer of Laboratory results from one health care organisation to another. The system would provide that any test result that is not reported as normal is flagged. Health information is fully encrypted before being transmitted from the NNSL to the maternity unit. MediBridge is an EDI (Electronic Data Interchange) communications environment, based on e-mail standards. There is no need to re-enter data into multiple systems. Once the information has arrived on the recipients workstation in the hospital, the information is decrypted and is available to view or integrate with their own information system.

## Training

The Medical Director and staff from the National Centre for Inherited Metabolic Disorders undertake a series of lectures on an ad hoc basis throughout Ireland for Public Health Nurses, and staff in maternity units. During the past eighteen months about one thousand nurses have attended these lectures. There is a need for standardised, regular in-service training for all staff involved in newborn screening testing.

It is the intention of the NNSL to place *The Practical Guide for Newborn Screening (2001)*[3] on the Web and the Medical Director is currently seeking financial support to progress this.

## Newborn Screening Cards

The European Newborn Screening Network has recommended S&S 903 as its paper of choice rather than S&S 2992, which is in current use in the Republic of Ireland. This paper is made to higher specification and as a consequence it will improve reproducibility. However, this change of paper will have no effect on the time needed to dry the sample.

The change of the collection paper used will provide an opportunity to redesign the NSC. Redesign of the NSC is necessary in order to capture more information and will facilitate the use of an optical card reader (OCR) to input patient demographic data into the computer and thus reduce transcription errors. There is currently no space for signed consent and ethnic group identifiers, nor is there a facility to allow the sample taker to retain a duplicate as proof of sample taking. The new cards will cost approximately three times (€60,000 per annum) the cost of the current cards.

## Storage of Newborn Screening Cards

NSCs are currently stored indefinitely at room temperature. This may lead to some deterioration in the quality of the sample. Best international practice suggests that NSCs should be stored at 40C. Currently there is no international agreed recommendation as to how long these cards should be stored for and the purposes for which they are stored. The NNSL has in storage all cards used for testing since approximately 1984. They are used in a linked fashion (identity of the infant known) by the NNSL, when requested by a pediatrician. This may happen approximately 5-10 times per annum, to re-check a diagnosis or to check a sibling of an infant with a diagnosis of a metabolic condition. They are used in an unlinked (anonymous) fashion by the NNSL to develop new assays for new tests and for research purposes following submissions of a strict protocol to and obtaining approval from the Children's University Hospital's Ethics Committee. This practice is currently under review.

## Weekend Testing

The NNSL is currently resourced to provide a routine service 5.5 days per week (Monday-Saturday morning). Assays set up on a Friday are read on Saturday mornings. An "on-call" weekend service is available to carry out testing for infants and children suspected of having a metabolic disease through the tertiary referral metabolic laboratory.

The incidence of Classical Galactosaemia in Traveller infants is 1:450 compared to 1:36,000 within the non-Traveller community. The NNSL protocol recommends that Beutler testing be carried out on all infants of Traveller parents and siblings of known cases to exclude Galactosaemia. This should be done immediately after birth. Given the current resources in the laboratory, the NNSL is not in a position to deliver a seven-day service. The Beutler assay is carried out on Saturday mornings and on Bank Holiday Mondays on samples received at the laboratory by 10 a.m. that morning. Mothers of Traveller infants and of siblings of known cases are advised to withhold Galactose containing feeds until the results of the test are declared normal.

CHAPTER 8

# Ethnic Minorities

## Introduction

*Best Health for Children. Developing a Partnership with Families*[4] states that the current system for surveillance and screening for children in Ireland is not equitable. The aim of this review is to provide an integrated, standardised yet flexible National Newborn Screening Programme (NNSP) for all groups and that best meets the needs of minority groups within reason.

In this chapter, ethnic minorities are discussed separately in recognition of the issues that must be overcome to ensure that the current NNSP remains a universal service for children in the Republic of Ireland. In this regard, consultation with representatives of ethnic groups and consultation with professionals working in the NNSP was arranged in order to explore possible barriers to uptake of newborn screening testing within these groups. From all the themes discussed language and mobility were viewed as the dominant barriers to uptake of newborn metabolic screening amongst ethnic minorities. There is little information available in an Irish context regarding ethnic minorities and newborn screening testing; hence the issues are examined as general barriers to health care and uptake of health services.

## Travellers

Consanguinity is common amongst the Irish Travelling community increasing the risk for autosomal recessive disorders, some of which may be inborn errors of metabolism[12]. As discussed previously the incidence of Galactosaemia among Travellers is high (1 in 450 as compared to 1 in 36,000 in the general population), justifying the need for special screening procedures i.e. the Beutler Test.

Thus, Irish Travellers are an “at risk” group that merit particular attention with regard to newborn screening testing2. However there is a low up-take of post-natal services amongst the Traveller community including newborn screening testing.

## Language

It is important that Travellers are aware of, and informed regarding newborn screening testing in order to increase their up-take rates. However it is estimated that up to 80% of Traveller adults have literacy problems[13] The involvement of Travellers in developing information has been identified, as the most effective method for ensuring that the information produced is clear and simple and can be understood by all. Visual health promotion campaigns that Travellers can self-identify with have had proven success in communicating health information to Travellers. A recent report recommended oral explanation regarding newborn screening testing and the Beutler test to Traveller mothers[14].

## Mobility

The nomadic lifestyle of Travellers is one tradition that has affected their access to screening. On discharge from hospital a Traveller mother and her new infant may move to a different health board area. This, in addition to the fact that Travellers do not have a fixed address makes it difficult for Public Health Nurses to trace the infant to report the results of the Beutler test or to take a newborn screening sample. A recent report has recommended that improving liaison procedures between maternity units and Designated Public Health Nurses for Travellers would improve the uptake of screening in this community[14].

## Refugees

It has been reported that applications for asylum in the Republic of Ireland have increased from 31 in 1991 to 10,938 in 2000[15]. In 2000, it is estimated that non-EU births accounted for 6% at the National Maternity Hospital, 15% at the Rotunda and 16% at the Coombe Women's Hospital. Although this figure undoubtedly includes non-EU nationals who are not seeking asylum, management at the hospitals are of the opinion that the majority are from the asylum seeking population.

While the majority of refugees and asylum seekers entering Ireland do not have the same high incidence of the five conditions screened for in Ireland, the NNSL recommends that all infants born to immigrant parents and all infants entering the country should have the screen performed. *A Practical Guide to Newborn Screening in Ireland (2001)*[3] also recommends that the decision to screen for other disorders should be taken by the local paediatrician, depending on the country of origin of the parents.

## Language

The countries of origin of the majority of asylum seekers are in Eastern Europe and Africa (See Figure 8.1). Not surprisingly communication difficulties for health service providers arise.

### Figure 8.1
### Top ten countries of origin for asylum seekers 2001

| *Country of Origin* | *No. of asylum seekers* |
|---|---|
| Nigeria | 3,461 |
| Romania | 1,348 |
| Moldova | 549 |
| Ukraine | 376 |
| Russia | 307 |
| Croatia | 292 |
| Democratic Rep. Congo | 281 |
| Lithuania | 246 |
| Poland | 242 |
| South Africa | 203 |

Literacy levels amongst refugees vary widely. Many Nigerian asylum seekers have third level education while it is estimated that approximately 80% of Roma (mainly women) have no English at all[16]. Health promotion material for ethnic minorities needs to be culturally sensitive and appropriate[17]. For health promotion material to be accessible to its audience, a visual campaign may be more successful than a written campaign targeting ethnic groups.

Simply providing a translator is not sufficient, the cultural context of the translation is also required. A recent report[18], developed in consultation with refugee and asylum seeking women, identified the lack of culturally sensitive translation facilities as the dominant health issue. A number of other problems arise around translators including impartiality, gender, trust and competence. In the case of small minority groups, the services of a telephone interpreter may be required, as translators may not be available on site. Such a service may be inadequate to explain symptoms and illness in a satisfactory manner. Given the technical language involved in newborn screening, this is likely to be particularly true.

## Mobility

Ethnic minorities in Ireland are a migratory population, which causes problems in provision of follow up services such as newborn screening testing in the community. Newly arriving refugees and asylum seekers are initially accommodated in reception centres. On the birth of an infant, a family may be entitled to seek independent or private housing thus changing address. It can be difficult for Public Health Nurses to locate such families as the address given by the hospital will be that of the reception centre.

Since April 2000, in response to a lack of accommodation in the Dublin area, asylum seekers arriving into the Irish State have routinely been resettled (dispersed) outside Dublin[19]. A small number of infants have been resettled prior to a sample being taken. This creates additional difficulties for Public Health Nurses as they may have to work across Health Board borders to ensure the sample is taken from the infant within the recommended time.

The movement of refugee and asylum seekers outside of Ireland following the birth of an infant is a further barrier to the uptake of newborn screening. A number of infants born in this country leave Ireland before they can be screened. Despite the efforts of professionals involved in the NNSP, these infants are untraceable and thus lost from the screening programme.

CHAPTER 9

# The Parents' Perspective

Qualitative research was carried out with a small group of mothers, who had recently had babies to establish;

- Their level of information concerning the "heel prick" test.
- Their views on the process, including consent and refusal.
- Any recommendations for improvement.

## Information

All mothers were aware of the test by virtue of having had previous babies, discussion with other mothers, friends and relatives.

Mothers are generally given information regarding the "heel prick" test before the procedure is performed or upon discharge from the hospital. The explanation and the detail of information given to parents varies. One mother stated that she had received no information from a health professional. Two mothers highlighted the fact that the period following birth is a very difficult time to take in information.

Mothers are offered verbal information or leaflets or sometimes a combination of both. For one woman the information leaflets made little sense.

The mothers interviewed stated that Midwives in hospitals provided information leaflets in contrast to community based Public Health Nurses, who gave verbal information as they were performing the test. Another mother commented that it can be difficult to concentrate on a discussion with a nurse while the baby is crying.

Some parents found the information too technical and too medical. However one acknowledged that it would be very difficult to simplify the information further while at the same time providing parents with accurate and detailed information.

Mothers' knowledge of the "heel prick" test varies. In general, their understanding is that the test is performed within 72 to 120 hours of birth, to detect abnormalities in the babies' blood. Most mums also knew that once detected, these conditions could be controlled using medication and dietary interventions. Mothers assume that the test is something that must be done for the good of their baby. They place their trust in the nurses and as a result do not ask questions about the test.

## Process

Policy in maternity hospitals varies in that mothers may be present or absent for the heel prick test. The majority of mothers interviewed, reported that their baby was taken away from them to the nursery for testing. One mother was happy to be absent from the actual sampling process as she felt her baby had undergone a number of other tests and did not want to see her baby upset any further. However most mothers would like to decide for themselves to be present or absent for the procedure. Mothers were quite ambivalent regarding their involvement in the testing Most used words and phrases such as "sit back", "on-looker" and "observer" to describe their role. Two mothers even felt that greater involvement on their part could impact negatively on their baby's test. While all of the respondents said that they could have asked questions, none did.

## Consent

In general parents appear to be happy consenting to the procedure. Most mothers reported that only verbal consent was sought and often in a very casual manner. One described it as similar to asking someone if they wanted a cup of tea. One woman felt that the nurse was not informative prior to seeking her consent. Other mothers were told that their babies were being brought for pre-discharge examination, during which testing would also be performed.
One mother could not remember if her written consent had been sought. She therefore thought it would be a good idea to have a written consent form with a duplicate for the Public Health Nurse and the parents as proof of the sample being taken. Another woman believed that written consent was necessary to protect the hospital against any future potential claims against the service.

## Refusal

None of the respondents had considered refusing the test. Two mothers assumed that the heel prick test was mandatory and that they had no right to refuse. Regardless of having an option they would still have the heel prick test performed on their baby. The resounding belief amongst the mothers interviewed is that the heel prick test is "good for baby". Parents are reassured and comforted by the fact that there are no perceived side affects associated with the heel prick test since it entails blood sampling rather than an injection. Many mothers spoke of their concern regarding the Vitamin K and MMR injections.

## Test Results

Parents are contacted only in the event of a "query positive" test result or if a repeat sample is required. Parents are not contacted in the event of a negative test result. Thus, they can only assume that "no news is good news"
Some parents had been advised that test results would be sent to their GP and would be available at the infant's 6-week check up. However mothers indicated a strong preference for receiving a written report of results.

## Improvements

The mothers made suggestions as to how the newborn metabolic screening programme service could be improved.

Parents want written information on all of the conditions tested for. They consider it important that information be given as to reason for testing and description of the test. They need written information in plain, simple language, with as little medical terminology as possible. This should be provided in the ante-natal period as mothers are emotionally vulnerable following the birth of the baby.

Some would like written consent to be requested prior to testing. It would be beneficial if a duplicate copy could be provided to parents.
Finally, respondents requested that parents should be informed of both positive and negative test results.

CHAPTER 10

# Discussion and Conclusions

A number of issues have been highlighted in this report that merit further discussion.

## Coverage

All respondents to the various questionnaires were confident that mechanisms were in place to ensure universal uptake of the newborn screening test. However, in the absence of a unique personal identifier number for all infants, there is at present no foolproof method for confirming this. The successful coverage is a measure of the commitment of all staff involved in the various stages of the screening programme.

## Responsibility

*The Report of Metabolic Disorders Working Group (1993)*[2] clearly states the responsibility of individuals in different settings but it does not make any recommendations regarding overall responsibility for the entire National Newborn Screening Programme (NNSP). Currently there is no mechanism in place to audit the programme, make evidence based decisions regarding inclusion of additional conditions in the screening programme, ensure that all research involving test samples is carried out in a fashion that complies with agreed ethical guidelines or up-date protocol as required.

In keeping with the recommendations of the 1993 Report, the majority of hospitals and community care areas nominate individuals as having responsibility for newborn screening testing. There is however a huge variation, especially amongst the hospitals regarding profession or grade to whom this role is designated. In essence hospital staff are concerned with ensuring the sample is taken.

## Protocol

*A Practical Guide to Newborn Screening in Ireland (2001)*[3] is acknowledged as being a very useful document. Service providers need clear policies and guidelines to reinforce and clarify their work practices. Although it is widely available not all personnel involved in the programme have a copy of these Guidelines from the National Newborn Screening Laboratory (NNSL).

Community care area personnel and domiciliary midwives encounter a wide range of difficulties in adhering to the protocol. In an attempt to ensure that testing is carried out within the time frame, as recommended by *A Practical Guide to Newborn Screening in Ireland (2001)*[3], Public Health Nurses endeavour to visit the mother and infant as early as possible in the working day. In order to ensure same day dispatch of the test sample, they must then personally access a post office if they are to obtain certificates of proof of postage. This may obviously be particularly problematic in a rural area. In addition, they frequently have problems in ensuring that the sample is totally dry prior to dispatch to the NNSL. This in turn leads to difficulty in processing the test at laboratory level, often leading to a request for repeat testing.

## Audit

A screening programme needs to be continually assessed and monitored to ensure it is doing more good than harm. There is currently no mechanism in place to review the quality of service being provided, and thus national, regional or local variations in service provision are not identified. Quality assurance and standardisation of the screening programme could be ascertained through

the development of a programme of audit and the development of performance indicators. This could be monitored by the Health Information and Quality Authority, to be established as set out in the *National Health Strategy Quality and Fairness*[20].

The National Health Strategy document Shaping a Healthier Future[21] published in 1994 challenged the health services to achieve measurable improvements in health and social gain and emphasised the need for comprehensive and good quality information. Furthermore the National Health Strategy 2001 and the Deloitte and Touche report both emphasised and recommended ICT development. An appropriate IT system is vital to the collection of information required for this. To collect data to assess the service, inform planning and enable evaluation of the effectiveness and efficiency of the screening programme, a standardised national IT system would be required.

## Weekend Testing

There is a specific timeframe within which samples should be collected i.e. after 72 hours and before 120 hours from birth. The reason for the specific timeframe is that some of the conditions, for example Galactosaemia and Maple Syrup Urine Disease, benefit from the earliest detection possible. Early detection may save the child's life and protect his/her intellect. If the sample is collected too late after birth some infants may present clinically before the results of the test are available.

Such a specific timeframe is not suitable for all conditions with the optimum time to screen for Homocystinuria being between day 7 and day 10. Evidence from audits of the NNSP and of European screening programmes suggests that screening programmes will not detect approximately one in five cases of Homocystinuria[22].

Most respondents to the questionnaires consider that weekend testing should be provided to ensure that testing is carried within the timeframe. Weekend testing is variable throughout the country. Practices for newborn screening testing at the weekend have developed in an uncoordinated manner, generally driven by resource constraints rather than having any regard for what is optimal. Parents may be requested to return to the maternity unit of birth or attend their local health centre with the infants for testing. In certain areas, parents will be advised that a Public Health Nurse will call to the home to perform the test.

The resources required to provide weekend testing are considerable; sample taking in the community and hospitals, timely transport of samples to laboratory and weekend laboratory cover.

Many respondents commented on the absence of a postal service during weekends or Bank Holiday periods. Even if the sample can be transported to the NNSL by another method of dispatch (ambulance, train, courier), no routine testing of samples is carried out at the NNSL over the period.

The NNSL is currently resourced to provide a routine service 5.5 days per week (Monday-Saturday morning). Assays set up on a Friday are read on Saturday mornings. An "on-call" weekend service is available to carry out testing for infants and children suspected of having a metabolic disease through the tertiary referral metabolic laboratory.

The incidence of Classical Galactosaemia in Traveller infants is 1:450 compared to 1:36,000 within the non-Traveller community. The NNSL protocol recommends that Beutler testing be carried out on all infants of Traveller parents and siblings of known cases to exclude Galactosaemia. This should be done immediately after birth. Given the current resources in the laboratory, the NNSL is not in a position to deliver a 7-day service. The Beutler assay is carried out on Saturday mornings

and on Bank Holiday Monday on samples received at the lab by 10 a.m. those mornings. Mothers of Traveller infants and of siblings of known cases are advised to withhold Galactose containing feeds until the results of the test are declared normal. In the case of a positive test result, the NNSL contacts the hospital directly.

## Transport of Samples

The NNSL has made a request to the sample takers to include a list of all babies whose samples have been included in any one batch sent to the laboratory. However, this request is not always complied with.

A range of transport systems is used in dispatch of samples from the hospitals or community to the NNSL. These include registered and ordinary post, courier, train, taxi or ambulance. Many of these arrangements pose practical problems for the staff using them and do not always guarantee timely delivery to the NNSL.

## Liaison

Newborn metabolic screening involves the collaboration of many parties, in the maternity units, the community and the NNSL, to provide a safe and comprehensive service. Relevant information needs to be communicated in a consistent, timely and effective manner.
The trend towards early discharge of infants into the community has heightened the need for a systematic Hospital-Community liaison system. The current liaison arrangements in place vary from one unit or area to the next, and a wide range of personnel carries out liaison. Given the nature of the role, it would be appropriate to nominate a person from a clerical background to communicate information between the hospital and the community. This person would report to the designated person with overall responsibility in the hospital for the NNSP.

Liaison procedures involving the NNSL also require standardisation. Hospitals and community care areas vary in their liaison practices with the NNSL. As a result the NNSL find it difficult to contact the relevant and responsible member of staff immediately when they have an urgent query for a hospital or a community care area. Nominating an individual with responsibility for co-ordinating the screening programme in hospitals and community care areas to liaise with the NNSL would make the liaison process more efficient. The liaison process is also made more difficult with no liaison officer to communicate with hospitals, community care areas or domiciliary midwives.

All queries are dealt with by the Medical Director or a Medical Scientist in the NNSL. The appointment of a liaison officer would assist in the flow of information to and from the NNSL.

## Reporting

Hospitals, community care areas and domiciliary midwives receive normal results, requests for repeat tests in the case of insufficient samples and query positive results, from the NNSL. Parents are more likely to be notified in the case of a positive result than a normal result by a hospital or a community care area. In some cases, results may not be communicated to the Public Health Nurse who takes the sample in the community. It is generally not policy for hospitals and community care areas to inform General Practitioners of any test results. Reporting a query positive result to parents must be done in a professional manner. It is acknowledged that the more information parents receive around the nature of their child's possible condition, the less anxiety they will feel during this stressful time. The NNSL has developed a protocol for reporting query positive results, including the information that will be required by the contact person to answer correctly any queries parents may have. The details are relayed from the NNSL, to an initial contact person,

generally situated in the maternity hospital of origin. This contact person may then ask the Public Health Nurse to locate and inform the parents of the result in the community. It falls to the Public Health Nurse to inform parents regarding positive results. It is obviously important that the Public Health Nurse herself is well informed in this regard.

## Recording

The importance of recording during various stages in the complex process of the newborn metabolic screening process cannot be over-emphasised. As it stands recording that the sample has been taken is the only standardised recording practice.
*The Report of Metabolic Disorders Working Group (1993)*[2] recommended a separate screening register be used for recording test information. While many hospitals and community care areas operate a register, it has not been taken on board universally and information continues to be recorded in a variety of files including the mother's files. Computerised records are in the minority with newborn screening test information being recorded manually by most hospitals and community care areas. "Medibridge", a pilot programme is currently assessing the feasibility of establishing a computerised data transfer system between the NNSL and maternity units, is currently under way at the Coombe Women's Hospital. The results of this pilot will determine future recommendations regarding electronic transfer of information between the hospital, community and laboratory interfaces. In February 2003, a joint application for funding was made to the ERHA by the Children's University Hospital, Temple Street, the Rotunda and the National Maternity Hospital to role out "Medibridge" in these hospitals also.

Checks to ensure that the information has been recorded and that no baby has been missed are carried out by the majority of hospitals and community care areas, but current practice could not be described as uniform or standardised.

## Consent

Obtaining consent before carrying out a clinical procedure represents fundamental good practice. To make an informed decision, sufficient information should be provided to enable parents to understand the process of and the need for newborn screening testing and thus to consent. The need for informed consent has been highlighted in the recent case heard in the High Court and subsequently appealed to the Supreme Court[8]. On this occasion, both courts upheld the parents' right to refuse newborn screening testing for their infant.

All respondents to the questionnaires stated that it is practice to inform parents about the purpose and the timing of newborn metabolic screening. In general, consent is sought at the time of sampling, with a minority seeking consent prior to sampling. Consent is commonly given orally, and in some situations, it is simply implied, (i.e. presence of a parent during testing is taken as implying consent for the procedure).

Written consent is not widely sought. Consent forms have been developed by certain individual units, but there is no nationally agreed consent form.

In the case of non-attendees and late attendees, consent should be obtained by hospital personnel in the post partum period.

Prior to written consent becoming routinely sought in the community consideration needs to be given to the liaison procedures around transferring the consent form there from the hospital. At present some hospitals transfer the NSC cards by giving the card to parents as they leave the hospital who in turn give it to the Public Health Nurse at the time of sample taking.

## Refusal

Parents have the right to refuse to allow their child to be screened. If a parent refuses, having been informed of the potential consequences of not screening, responsibility for the possible adverse consequences of their decision rests with the parents. The management of the maternity unit has no option other than to comply with the parent(s)' wishes to decline the screening test. As a result *The Practical Guide to Newborn Screening (2001)*[3] recommends that once parents have been fully informed of the consequences of their decision, they should be requested to sign a refusal form. A copy of this form should then be sent to the NNSL. Refusal forms are used in a small number of community care areas and hospitals but the contents of these forms vary. In an effort to standardise the process of a refusal, the NNSL has developed a draft refusal form (see appendix 3).

## Parent Information

The Department of Health and Children currently publishes health promotion material on newborn screening testing but the general consensus is that this information requires up-dating, particularly in view of the need for culturally appropriate material for an increasingly multi-cultural population. Consideration must also be given to where parents can get this information. Antenatal classes, GP surgeries, maternity wards and health centres are appropriate points to distribute such information to expectant parents. Currently it appears that the norm is for parents to receive information at the time of sample collection. This may be given verbally, but information leaflets regarding newborn screening testing are available.

Both the hospital and community sectors are attempting to cater for the specific needs of ethnic minorities. It is widely recognised that lack of appropriate and meaningful information is one of the barriers to uptake of preventive services by ethnic minority groups. Ethnic minorities have special information needs which must be recognised and catered for. The present information leaflets are inadequate to meet cultural diversity. Visual messages through videos and posters are seen as the most appropriate way to communicate information to ethnic minorities. Additionally the issue of poor literacy is also overcome through visual information. Culturally acceptable translation services are required to provide an equitable health service however a number of respondents stated that Translators are not universally available for newborn screening testing. It is accepted that health promotion materials developed in consultation with minority groups are more successful in conveying health information.

## Travellers

Ethnicity is highly relevant with respect to newborn screening testing, as different ethnic groups will experience different incidences of the five disorders tested for to those experienced by the population as a whole. As outlined previously, Irish Travellers experience a higher rate of classical Galactosaemia than that of the population at large. The introduction of an ethnic group identifier would alert medical and nursing personnel to the need for additional screening for Travellers and appropriate advice regarding infant feeding pending newborn screening test results.

## Refugees and Asylum Seekers

Many of the ethnic groups arriving to the Republic of Ireland as refugee and asylum seekers do not experience the same high incidence of the five conditions included in the NNSP. However, it is more cost-effective to offer it to all infants in a universal fashion rather than to exclude certain ethnic groups from routine testing. Additionally, infants from certain ethnic groups or from certain countries of origin may require screening for additional disorders not tested for universally in the Republic of Ireland. *A Practical Guide to Newborn Screening in Ireland (2001)*[3] recommends that individual paediatricians should determine if infants from certain ethnic groups require specific

additional testing. Screening for Haemoglobinopathies has commenced in an ad hoc fashion in some laboratories around the country. This is not cost effective and not in accordance with established best practice, as numbers screened at each centre are low.
Screening programmes vary amongst countries, and children arriving into Ireland may not have been screened for the five disorders included in the Irish programme or indeed for any inherited and metabolic disorders in their country of origin. PKU, Homocysinuria and Congenital Hypothroidism may present late with minimal clinical symptoms.

## Training

All personnel involved in the NNSP need to have an understanding of its objectives to ensure their full compliance with *A Practical Guide to Newborn Screening in Ireland (2001)*[3] Managers and clerical staff need training in documentation and filing of records. Nurses, in both hospital and community settings require updated information regarding the conditions being screened for, and also, specific training in sample taking and handling. All groups of personnel consulted during this review identified the need for ongoing training. There is currently nothing in place to ensure uniform training standards are adopted throughout the country. Training in "newborn screening testing" is included in the curricula for midwives and Public Health Nurses. Provision of in-service training however, is very uneven. Staff from the National Centre for Inherited Metabolic Disorders are happy to organise lectures, but do so only at the request of management.

## Equipment

*A Practical Guide to Newborn Screening in Ireland (2001)*[3] provides a list of equipment needed to take a sample for newborn screening testing and based on this, standardised equipment is available to sample takers in all community care areas. Public Health Nurses from ERHA have suggested that a newborn screening testing pack be developed to facilitate correct sample taking. Public Health Nurses and domiciliary midwives taking samples in the community face the significant challenge of drying samples, prior to dispatch. Frequently the NSC is left exposed in the sample takers car, in an effort to dry it prior to postage or dispatch. Inadequate drying may lead to the need for repeat testing. In response to the request made by Public Health Nurses, the NNSL has agreed to develop a standard newborn screening testing pack which can be used as a prototype by community care areas in assembling their own packs.

## Newborn Screening Card

It is envisaged that the paper used in the collection systems of European Metabolic screening programmes will be standardised to type S&S903 in the future. In Ireland this would mean changing the collection paper currently used, thus providing an opportunity for re-design of the NSC. It is therefore timely to consider perceived problems with the existing card and its storage.

The majority of respondents to the questionnaires suggested that the current NSC requires up-dating and re-design for a variety of reasons;
The card is too small to facilitate ease of completion by sample takers. Failure to include all details often leads to the need for additional and time wasting input from staff at the NNSL.

Community based nurses in particular experience difficulty in drying the cards fully, prior to their placement in the approved envelope. As previously discussed, the sample may as a result be unsatisfactory, necessitating a repeat sample and test.

Respondents would also like to record additional information such as mother and child's surname, place of birth etc.

Respondents have also suggested that the card could be produced in duplicate form, to allow the sample taker to document details re sample taking in the infant's chart.
Others commented on the desirability of including a unique client identifier number (such as the PPSN- Personal Public Services Number), an ethnic identifier and the parent's signature on the NSC.

Currently there is no unique client identifier issued at the birth of a child. Rather the existing practice is that each hospital of birth assigns a chart or hospital number to each baby, which is unique only for that establishment. This practice excludes domiciliary births.

The Personal Public Service Number (PPSN) was introduced in the 1998 Social Welfare Act as the unique personal identifier for transactions between individuals and Government Departments and other agencies specified in the Social Welfare Acts. Bodies authorised to use the PPSN include health boards and voluntary hospital including The Children's, Hospital, Temple Street. The PPSN is used as a unique client identifier by The Department of Health and Children Immunisation Programmes and Irish Cervical Screening and Breast-Check Programmes. It is particularly useful where there may be a number of people with similar or identical names and/ or addresses. The PPSN represents an option for the neonatal screening programme. However PPSNs are assigned at the time of application for Child Benefit rather than at birth. Despite future plans to allocate the PPSN earlier (at the time of registration of the birth) the PPSN is not likely to be an option for the newborn screening programme in the near future.

From the NNSL point of view there are also problems with the storage of NSCs. There is no international consensus around recommended duration of retention of NSCs. The NNSL has in storage all cards used in testing since approximately 1984. Additionally these cards are stored at room temperature, while best international practice suggests that NSCs should be stored at 4ºC.

It was also highlighted that cards are currently being retained without the full knowledge of parents and it is suggested that parental consent be sought for retention of card in the future. Seeking informed consent would require parents knowing about the benefits of using stored cards in a linked (i.e. identity of infant known) and unlinked (anonymous) fashion.

## Funding

Funding for the NNSL is allocated through the ERHA and is included within the Children's University Hospital annual allocation. It is not specifically identified and consequently not ring-fenced. The NNSL although providing a national programme is thus competing for resources with other services within the hospital itself. This is obviously unsatisfactory for both the Hospital and the NNSL.
Current equipment used within the NNSL is outdated. The bacterial inhibition assays do not conform to Good Laboratory Practice nor would they withstand the rigors of Laboratory Accreditation procedures. Tandem Mass Spectrometry is the recognised method of choice, providing a rapid, sensitive quantitative method. Tandem Mass Spectrometry has many advantages including higher sensitivity and specificity than current methods used. A formal request was made to the Department of Health and Children in 1999 for the implementation of Tandem mass Spectrometry. However, to date, the NNSL has been unable to access funding. In order to improve the current system there are a number of initiatives taking place.

The computer system used by the NNSL does not have dedicated screening software and as a consequence, recovery of data is limited. Additionally the ultimate aim of redesigning the NSC is to use an optical card reader to input patient demographic data and thus reduce transcription errors. The current computer system is not compatible with an optical card reader.

There are a number of developments that will also involve the modernisation of the current system. A pilot programme involving the implementation of "Medibridge" is currently assessing the feasibility of establishing a computerised data transfer system between the NNSL and the Coombe Women's Hospital. The results of this pilot will determine future recommendations regarding electronic transfer of information between the hospital and maternity units, and community care areas.

Approximately 58,000 specimens are processed each year in the NNSL. This throughput is in accordance with the optimum size of a newborn screening laboratory, as recommended by the Directors of UK Laboratories. However there are currently too few medical scientists employed to deal with this workload, nor is there a Quality manager employed to oversee the testing process at the NNSL.

Staff in the NNSL are currently performing duties beyond their roles. There is currently no Clinical liaison officer in the NNSL appointed to liaise with maternity units and community care areas. At present, this important but time consuming function is undertaken by senior technical staff and by the Medical Director.

CHAPTER 11

# Recommendations

## GENERAL RECOMMENDATIONS

### Responsibility

- Overall responsibility - An Executive Committee should be established with a brief to oversee and to have ultimate responsibility for the programme. Since the screening programme is national, it should report to the Department of Health and Children.

- This committee should audit the programme on an annual basis, make evidence based decisions regarding inclusion of additional conditions in the screening programme and ensure that all research involving test samples is carried out in a fashion that complies with agreed ethical guidelines.

- Representation on this committee should include:
  Department of Health and Children
  CEO, Health Boards
  Medical Director, National Newborn Screening Laboratory (NNSL)
  Metabolic Unit, The Children's University Hospital
  Director of Public Health Nursing
  Director of Nursing, Maternity Hospital
  Faculty of Paediatrics
  Faculty of Public Health Medicine
  Institute of Obstetrics and Gynaecologists
  Parent Patients' Organisation

- The working group endorses the recommendations of the Metabolic Disorders Working Group (1993) regarding;

**Responsibility of Health Boards:** 'The ultimate responsibility for ensuring that all infants are screened rests with the Health Boards. The Health Boards are responsible for ensuring that the tests are carried out in accordance with agreed protocols.'

**Responsibility of Hospitals:** 'Hospitals are responsible for ensuring that all infants born in hospital are screened for metabolic disorders. If the test is not performed before discharge from hospital, the hospital is responsible for ensuring that the child is screened by appointment at the hospital or that the Director of Public Health Nursing is informed that the infant has been discharged prior to the test being carried out. If no results are received from the National Screening Laboratory by the hospital, the hospital is responsible for ensuring that omissions are notified to the Directors of Public Health Nursing.'

**Responsibility of health board community care services:** 'If the test is carried out in the community, the recording and follow- up of the case is the responsibility of the Directors of Public Health Nursing.'

**Responsibility of general practitioners and midwives:** 'The midwife and the general practitioner performing home deliveries are responsible for performing the test.'

## Funding for Metabolic Screening

- The National Newborn Screening Laboratory for Inherited Metabolic Disorders should receive an independent "Ring-fenced" budget separate from that of The Children's University Hospital, Temple Street.

- Funding will also be required to provide additional resources as set out in the report;
  Tandem Mass Spectrometry
  IT specific to the laboratory
  IT support
  Additional staff in the NNSL, hospital and community sectors

- Each Health Board should review the NNSL revised protocol and determine its own requirements accordingly.

## Audit

- A review of newborn metabolic screening should be undertaken in each Health Board.

- This should include an audit of current practice in respect of both hospital and domiciliary births.

- Results from this review should be reported to the Executive Committee.

- A system of on going audit should be incorporated into the programme to ensure quality assurance and the delivery of a standardised screening programme.

- Registers containing data regarding metabolic screening should be computerised. Once Medibridge, has been evaluated and refined, consideration should be given to the planning of a national roll-out.

- A standardised national IT system, that will allow for transfer of data between the maternity units, the NNSL and the community sectors, must be developed. This will aid audit and production of performance indicators.

- The CEOs of the 10 health boards have commissioned the development of suite of performance indicators. Data regarding the NNSP should be collected by each health board;

- Total number of infants screened.

- Number of infants screened in the hospital.

- Number of infants screened in the community.

- Number of infants screened by the domiciliary midwives.

- Total number of infants tested within the recommended timeframe.

- Total number of positive results.

Collection of this data will not be feasible with the current manual system.

## Protocol

- *A Practical Guide to Newborn Screening in Ireland*[3] developed by the NNSL should be updated to incorporate the accepted recommendations and adopted as a nationally agreed protocol.

- This protocol should be available to all personnel, involved in *The National Newborn Screening Programme (NNSP).*

- All community care area and hospital protocols should be closely based on *A Practical Guide to Newborn Screening in Ireland*[3].

## Weekend Testing

- Samples for newborn screening should be collected when an infant is aged 72-120 hours, in keeping with the *A Practical Guide to Newborn Screening in Ireland*[3].

- The current yield from weekend testing does not however justify the investment required to provide additional resources for weekend testing. Thus considerable emphasis should be placed on provision of an optimum 5-day week service, with every effort made to ensure that a child is tested within the recommended time frame.

- Diagnostic Beutler Testing is performed in the NNSL on Saturday mornings to check for Galactosemia. Professional staff must ensure that appropriate dietary advice is given to mothers of Traveller infants and siblings of known cases until the results of the Beutler test are available. Specifically, these mothers must be advised to withhold Galactose containing foods and to defer breast-feeding pending normal Beutler test results.

- Consideration should be given to all of the above recommendations in the development of the Health Boards protocol.

## Transport of Samples

- Each Health Board should review its transport systems to ensure that samples are dispatched in a cost-effective and timely fashion.

- HeBE, through the National Health Care Materials Management Board should be requested to assess the feasibility of developing a dedicated national courier service for the NNSP.

## Recording

- All newborn screening tests taken must be documented by the sample taker at time of sample collection. This will facilitate coverage and audit of the programme.

- A designated person should have specific responsibility for recording, up-dating and checking the screening register. The register should be checked weekly to ensure that no infant has been missed.

## Reporting of Results

- Parents should be informed of appropriate test results including normal results. Details of results may be recorded in the Personal Health Records.

- Normal Results - Since the Public Health Nurse is likely to have ongoing contact with the mother, she should inform parents of normal results. It will therefore be necessary to ensure that the relevant Public Health Nurse is informed of the result by the designated person at community care area level.

- Requests for Repeat Sampling - The liaison person at the maternity unit should either contact the parents and arrange for the infant to attend the maternity unit for a repeat sample or contact the Director of Public Health Nursing. The Director of Public Health Nursing must then arrange for a Public Health Nurse to visit the parents and take a further sample.

- Query Positive results - The response to query positive results should be immediate and direct.

- Positive Results - The procedures for reporting requests for positive results as laid out in the NNSL Practical Guidelines should be followed.

- All individuals involved in reporting positive results to parents should receive training regarding the procedure for reporting results as developed by the NNSL.

- A check-list of information should be available to Public Health Nurses who may be asked to locate and inform parents of possible positive test results.

## Parent Information

- The executive committee should be responsible for the development and updating of Health Promotion material. Health Promotion materials for national use should include leaflets, videos and posters. They should be readily available in antenatal classes, GP surgeries, maternity wards and health centres.

- Each maternity unit must ensure that information regarding screening for Inherited Metabolic Disorders, both verbal and written, is provided for parents during the antenatal period.

- Ethnic minorities have special information needs, which must be recognised and catered for. In order to ensure that health promotion materials are developed in a culturally sensitive and meaningful fashion consultation should occur with Traveller and refugee support organisations.

- The Working Group endorse the recommendations from the report The Maternity Care Needs of Refugee and Asylum-seeking Women[13] regarding:

  'A standard translation service should be provided by the health authorities and not dependent on an NGO sector unless the government contracted the work to them on a formal basis.'

  'The establishment of a 24 hour telephone language interpretation service similar to Language Line, the London based service is vital.'

  Provision of 'Race awareness training programmes'.

- Appropriately trained and resourced Public Health Nurses should be employed in community care areas, with a brief to act as a support for parents from ethnic minority groups.

## Consent

- Consent for newborn screening testing must be sought on all occasions and must be informed and written.
- All sample takers should use a standardised national consent form.
- Hospital personnel should ideally obtain written consent during the antenatal period.
- In the case of non-attendees and late attendees at antenatal clinics, consent must be obtained by hospital personnel, in the post partum period or by the public health nurse.

## Refusal Form

- A refusal form should be developed by the NNSL for use by each Health Board. (see appendix 3)
- Implications of not having an infant tested should be clearly outlined in the form.
- Refusal forms should be widely available to all community and hospital staff involved in taking a sample.
- Every effort should be made by personnel involved in sample taking to encourage uptake. Parents should receive both verbal and written information. In the case of a refusal a standardised refusal form must be signed preferably by both parents and witnessed after they have been given both written and verbal information about the screening programme.
- A copy of the refusal form must be sent to the NNSL and to the infant's General Practitioner to advise them of the refusal.

## Travellers

- Ethnic group identifiers should be developed and included on the Newborn Screening Card (NSC) to facilitate timely screening of high risk infants.
- The working group endorses recommendations from the report *Traveller Health A National Strategy*[11] that, 'The need for special tests such as the Guthrie test and Beutler test will be adequately explained to Traveller mothers in the ante-natal period. Mothers will be supported and encouraged to stay for an appropriate period of time in hospital following birth so that the full range of post- natal services is availed of.'
  'Vigorous efforts will be made to ensure that Traveller babies receive the full range of neonatal metabolic screening. If this involves longer stays in hospital post- partum to ensure that babies are not lost to follow up, this will be considered'.

## Refugees

- Ethnic group identifiers should be developed and included on the Newborn Screening Card (NSC) to facilitate timely screening of high risk infants.
- Cases of metabolic disease have occasionally been detected in older children. Following best practice guidelines, all children up to the age of 16 years, irrespective of ethnicity, arriving in Ireland should have the full screen for inherited and metabolic disorders performed. Where feasible this should take place during general health screening for Refugees and Asylum Seekers following entry into the country.

- Individual Paediatricians should continue to determine if infants from certain ethnic groups require specific additional testing.

- The NNSL should be resourced to provide a Haemoglobinopathy screening and a diagnostic service on a national basis.

## Training

- It is the responsibility of each Health Board to ensure that all personnel involved in the NNSP in their region receive appropriate training.

- The working group endorses the recommendation of the report, *The Scope and Midwifery and Nursing Practice*[23] that, "Continuing professional development following registration is essential for nurses and midwives in order that they can acquire new knowledge and competence that will enable them to practice effectively in an ever-changing health care system. The nurse or midwife has responsibility to develop himself or herself as a professional".

- The training for Public Health Nurses regarding the NNSP should be incorporated into the training programmes for nurses in child health surveillance, which is being developed following recommendations in a BHFC report, *Training of Doctors and Public Health Nurses in Child Health Surveillance*[24].

# SECTORAL RECOMMENDATIONS

## HOSPITAL

### Responsibility

- A designated person should be nominated in each hospital or maternity unit as having overall responsibility for the NNSP. The NNSL and relevant community care areas must be informed immediately regarding any change in designated personnel.

### Liaison

- A designated liaison person with responsibility for all liaison with the NNSL and the community care areas should be nominated in each maternity unit. Hospital management will nominate the designated liaison person. The nomination process will be regulated through Health Board protocol and auditing.

### Training

- Training regarding the NNSP should be incorporated into the curriculum for student midwives.

- The person with designated responsibility for the newborn screening programme in each maternity unit, should ensure that all relevant personnel employed in the unit receive appropriate in-service training.

- All units should use the same guidelines and the information should be easy to access.

## COMMUNITY

### Responsibility

- A designated person possibly the Assistant Director of Public Health Nursing should be nominated in the community as having overall responsibility for the NNSP.

### Liaison

- A designated Public Health Nurse with the appropriate clerical support should be nominated in each community care area with responsibility for liaison with the NNSL and the maternity units.

- Public Health Nurses should be advised regarding outcomes of newborn screening testing for all children in their respective patches.

- Electronic communication and fax machines should be available in all health centres to facilitate improved communication between the hospital and community sectors.

### Reporting Results

- Each Public Health Nurse should have access to a detailed information pack prior to advising parents of possible positive test results.

### Training

- Training regarding the NNSP should be incorporated into the curriculum for student Public Health Nurses.

- Directors of Public Health Nursing should ensure that all Public Health Nurses employed in their community care area receive regular in-service training on the NNSP.

- A training programme for midwives and public health nurses should be developed. Training will focus on and address issues specific to the NNSP in the Republic of Ireland. The training may be delivered by the NNSL subject to appropriate resource provision.

- Consideration should be given to joint training of Midwives and Public Health Nurses.

- In-service training should be incorporated into the planned training scheme for doctors and public health nurses involved in child health surveillance[24].

### Equipment

- All community-based nurses involved in sampling should be provided with a NNSP sample takers pack as developed by the NNSL.

## DOMICILIARY MIDWIVES

### Protocol

- All domiciliary midwives should have a copy of the NNSL guidelines and adhere to its recommendations.

### Training

- Domiciliary midwives should receive regular in-service training on the NNSP.

- The Newborn Metabolic Screening Working Group endorses the recommendation of the report, *The Scope and Midwifery and Nursing Practice*[23] that "Continuing professional development following registration is essential for nurses and midwives in order that they can acquire new knowledge and competence that will enable them to practice effectively in an ever-changing health care system. The nurse or midwife has responsibility to develop himself or herself as a professional".

### Equipment

- All domiciliary midwives involved in sampling should be provided with a NNSP sample takers pack as developed by the NNSL.

### Reporting

- Domiciliary midwives should inform the Director of Public Health Nursing that the test has been taken.

- Test results should be sent to both the Domiciliary midwife and the Director of Public Health Nursing.

## NNSL

### Equipment

- National screening should continue to be centralised at the NNSL.

- Tandem Mass Spectrometry should be installed as a matter of urgency to ensure compliance with established Good Laboratory Practice and with accreditation procedures, replacing the BIA (bacterial inhibition assay) for screening for PKU, MSUD and homocystinuria.

- The laboratory information system should in due course be replaced by a Laboratory Information System capable of incorporating an optical card reader to input patient demographic data.

### Staff

- In order to enhance service provision the NNSL should be funded to appoint:
  a Liaison officer
  a Quality manager
  additional medical scientist

### Card

- The collection paper used should change from S&S 2992 to S&S 903. This paper is made to higher specification and as a consequence it will improve reproducibility. The change of the collection paper used will provide an opportunity to redesign the NSC. Redesign of the NSC is necessary in order to capture more information and will facilitate the use of an optical card reader (OCR) to input patient demographic data and thus reduce transcription errors.

- The Newborn Screening Cards (NSCs) should be redesigned to incorporate the following;
  Space for signed consent
  P.I.N./PPSN
  Ethnic group identifier

- The cards should be designed in duplicate form to allow the sample taker retain a duplicate, as evidence of having taken the sample.

- NSCs should be stored at 40ºC.

- Cards should be stored in keeping with the expected Guidelines from the Organ Retention Tribunal.

### Weekend Testing

- The NNSL must be closed for no period greater than 48 hours.

- The NNSL must be funded to provide a diagnostic service on Bank Holiday Mondays and over long weekends such as Easter and at Christmas.

## Liaison

- A designated liaison person with responsibility for all liaisons with maternity units, community care areas and Domiciliary Midwives must be appointed in the NNSL.

- This designated liaison person should have responsibility to notify the relevant General Practitioner of a positive result.

## Requests for repeat sampling

- All requests for repeat sampling should be communicated as speedily as possible. Requests should be faxed to the liaison person at the maternity hospital where the child was born.

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# Appendix 1

HAZARD ANALYSIS CRITICAL CONTROL POINT APPROACH

### 1a.Critical Control Point
Birth - Hospital

### 1b.Critical Control Point
Birth - Domiciliary
Control
Protocol
Notification in birth register

### 2a.Critical Control Point
Infant 72-120 hours old-in hospital.
Control
Protocol
Request for Guthrie

### 2b.Critical Control Point
Infant 72-120 hours old-in community.
Control
Protocol
Request for Guthrie

### 3.Critical Control Point
Taking of sample
Control
Protocol
Training of staff
Appropriate equipment.

### 4.Critical Control Point
Preparation of sample for dispatch
Control
Protocol
Training of staff
Appropriate equipment.

### 5.Critical Control Point
Dispatch of sample
Control
Protocol
Training of staff

### 6.Critical Control Point
Arrival of sample at The Children's University Hospital Temple Street Laboratory.
Control
Protocol
Receipt by clerical officer.

### 7.Critical Control Point
Analysis of sample.
Control
Protocol
Staff training.
Appropriate and well maintained equipment.

### 8a.Critical Control Point
Communication of results of test -normal
Control
Protocol
Clerical officer.

### 8a.1.Critical Control Point
Results received by referring agency
Control
Protocol
Clerical officer.
Birth Register

### 8a.2.Critical Control Point
Results communicated to parent.
Control
Details noted in infant's chart

### 8b.Critical Control Point
Communication of results of test -Positive
Control
Protocol
Clerical officer.
Director of TSH Lab.
Paediatrician.

### 8b.1.Critical Control Point
Results received by referring agency
Control
Protocol
Clerical officer.

## 8b.2.Critical Control Point

Results communicated to parent.
Referral for retest and medical follow-up.
Control
Protocol
Clerical officer.
Director of TSH Lab.
Paediatrician.
Details noted in infant's chart

## 8c.Critical Control Point

Communication of results of test –Unsatisfactory, needs to be repeated
Control
Protocol
Clerical officer.
Director of TSH Lab.

## 9c.1.Critical Control Point

Submission of new sample to lab. Steps from No. 4.

# Appendix 2

## FUNDING COSTS FOR NATIONAL NEWBORN SCREENING LABORATORY

| | Cost in Euro | | |
|---|---|---|---|
| | **2003** | **2004** | **2005** |
| ONGOING COSTS | | | |
| Pay costs (salaries)<br>2 WTE senior medical laboratory scientists<br>2 WTE basic medical laboratory scientists<br>1 WTE Grade IV administrative officer<br>1 WTE Grade III administrative officer<br>0.3 WTE Director<br>0.25 WTE Laboratory Manager<br>0.16 WTE Chief Medical Laboratory Scientist | €295,000 | €309,000 | €324,000 |
| Non-pay (reagents, QC, Maintenance contracts etc) | €190,000 | €197,000 | €205,000 |
| New cards | €15,000 | €75,000 | €78,000 |
| COSTS TO IMPLEMENT RECOMMENDATIONS FROM REPORT | | | |
| Tandem MSMS | | | |
| Capital expenditure | | €540,000 | |
| Recurring<br>1 WTE Senior Medical laboratory scientist<br>Service agreement & consumables | | | €104,000 |
| Liaison staff 1WTE CNM II | | | €50,000 |
| Extension of IT transfer system and maintenance | | | €10,000 |
| Training for Public Health Nurses and midwives | | | €20,000 |
| Attendance of staff at international meetings | | | €5,000 |
| Board meetings | | | €2,000 |
| TOTAL | €500,000 | €1,121,000 | €798,000 |

# Appendix 3

## NATIONAL NEWBORN SCREENING PROGRAMME FOR INHERITED METABOLIC DISORDERS DRAFT REFUSAL FORM

*Baby's Surname* *Baby's First Name*

*Date of Birth* *Hospital Number*

*Hospital of Birth* *Ward/ Community Care Area*

*Baby's Home Address*

*Mother's Surname* *Mother's First Name*

We _____, being the parents / guardians of Baby _____ refuse to allow the Newborn Screening Test (the Heel-prick test) for inherited metabolic disorders to be carried out on our baby. We have read the Department of Health and Children's leaflet on Newborn Screening; this has also been explained to us. We fully understand the gravity of the decision that we are taking by not allowing our baby to be tested. We understand that the medical consequences of not detecting or treating one of these disorders, should our baby have one, might result in severe mental or physical handicap necessitating possible institutional care or in premature death.

*Signed*

*Full Names* *Date*
(BLOCK CAPITALS)

*Witnessed by* *Date*

*Rank* *Community Care Area*

**Official use only.** Copies of this completed form, duly signed by both parties, should be forwarded to the Director of Public Health Nursing, the National Newborn Screening Laboratory and to the Baby's General Practitioner.

| | **Director of Public Health Nursing** | **National Newborn Screening Laboratory** | **General Practitioner** |
|---|---|---|---|
| *Name* | | National Newborn Screening Laboratory | |
| *Address* | | The Children's University Hospital Temple Street, Dublin 1 | |
| *Date Sent* | | | |

The Health Boards Executive

*Working Together for Health*

**Programme of Action for Children**


DUBLIN
Units 8/9
Manor Street Business Park
Manor Street
Dublin 7
Tel +353 (0) 1 868 2942
Fax +353 (0) 1 868 2943
Email pac.dublin@hebe.ie

LIMERICK
57 O'Connell Street
Limerick
Tel +353 (0) 61 310 437
Fax +353 (0) 61 310 612
Email pac.limerick@hebe.ie

SLIGO
3rd Floor
Bridgewater House
Sligo
Tel +353 (0) 71 917 4780
Fax +353 (0) 71 913 8335
Email pac.sligo@hebe.ie